

MSUD NEWSLETTER

Published by the Maple Syrup Urine Disease Family Support Group • Volume 25, Number 2 • Fall • 2008



What I learned On My Summer Vacation

Karen Dolins, EdD, RD, CDN
Newsletter Editor

Our summer began with a trip to Columbus, Ohio for the MSUD Symposium. Sadly, Hannah became ill almost as soon as we arrived, but our MSUD family jumped into action. Sandy Bulcher gave us the phone number to call at the hospital, and her husband Dave played ambulance driver getting us to the ER in no time. Fortunately, after a few hours of IV fluids, Hannah was able to leave. While we missed the day's sessions, we had a great time playing MSUD Jeopardy when we returned.

Fast forwarding to August, our summer ended with a road trip from New York to Chicago. Along the way we spent a day and night with the Brubacher Family in Goshen, Indiana. As most of you know, the Brubachers were among the first to have a child diagnosed with MSUD all the way back in 1965. Listening to them tell how they learned how to treat Monte, their first-born son, and Shayla, essentially through trial and error, makes what our family went through look like a walk in the park. Monte sadly succumbed to brain edema at age 9. Shayla is a testament to her parent's never-ending devotion and perseverance. Their collective wisdom leaves me in awe.

After spending a lovely day swimming in the Brubacher's home-made pond and playing croquette on their home-made field, we drove on to Chicago. There we met up with Dylan Mudrick



Medical Management of MSUD

Gerard T. Berry, M.D.
Division of Genetics
Children's Hospital Boston

Symposium 08

MSUD is a disease that greatly illustrates not only chemical individuality, but also personalized nutrition. The beauty is in its simplicity and adherence to the principles of intermediary metabolism and modern nutrition. Chronic care of the MSUD patient requires an understanding of the branched-chain amino acid (BCAA) nutritional requirements of patients of varying ages with an appreciation of the concept of biochemical individuality.

Normal growth and development and control of branched-chain amino acid levels is enabled by the use of a medical food providing a mixture of all essential nutrients including all but the branched-chain amino acids combined with foods or solutions containing just the right amount of leucine, isoleucine and valine to satisfy age-dependent requirements for protein synthesis. The ability to measure BCAA levels with relatively rapid turn-around time is also required.

(Dr. Berry cont. on page 6)

(Editor's Message cont. on page 8)

Inside This Issue:



Article, Medical Management	cover, 6	Article, Nutritional Management	9, 10, 11
Message from Editor	cover, 8	Article, Transition Into Adulthood	12
Symposium Scrapbook	2	Diet Wise	13, 14
Symposium 2008 Overview	3	Swimming by Hannah Dolins	15
Article, MSUD After You Grow Up	4	Article, Busy On The Hill	16
Member Story, Delecta Li	5	Make-A-Wish	17, 18
Members Keeping in Touch	5	Article, Beyond Low Protein	19
Article, Newborn Screening	7, 8	Contacts	back page



Symposium 2008 Scrapbook



Symposium Overview

Sandy Bulcher

Director, MSUD Family Support Group and 2008 Symposium Coordinator

MSUD Symposium 2008 was held at the Embassy Suites in Columbus, Ohio on June 26-28th. Approximately 210 people attended the three day event. MSUD families from 20 of the 50 United States were present. In addition, MSUD families from Canada, Philippines, Ecuador, India, United Arab Emirates, and England attended the conference. Sixty individuals with MSUD ranging in age from 2-39 were present.

A reception was held Thursday evening, in the beautiful atrium of the Embassy Suites. During that time, families enjoyed renewing old friendships and meeting new families.

Friday was a full day with many topics and speakers. We were honored to have Dr. Gerard Berry from Boston, MA participate in the conference. He spoke on medical management of MSUD. Following Dr. Berry, Indira Mehta of San Francisco informed us of a new study that she is conducting regarding young adults with MSUD. Next, Rick Finkel of Applied Nutrition shared information about new products available through his company. Just before lunch, Dr. Kim McBride of Nationwide Children's Hospital Columbus, Ohio covered issues related to aging and chronic disease. Lunch was served in the hotel atrium and included many delicious low protein choices. During the lunch break, families and professionals had the opportunity to view Amy Zimmerman's transplant DVD.

The afternoon began with Lynn Seward, RN from University of Pittsburgh Transplantation Institute enlightening us on commonly asked transplant questions. Families and professionals then had an opportunity to visit the vendors that participated in the conference, including Abbott Labs, Cambrooke Foods, Nutricia North America, Vitaflo, and Applied Nutrition. Karen McHugh of Nationwide Children's Hospital Columbus, Ohio then reminded the group that siblings of those with chronic illness need support also. The sessions concluded with the professional panel. Dr. Gerard Berry, Dr. George Mazariegos, Lynn Seward RN, and Sandy vanCalcar RD answered questions posed by parents. The session proved to be very interesting and educational for families.

The day concluded with a group photo of those with MSUD. It was challenging to get 60 MSUD children and adults to stand still while many parents took pictures, but it was very worthwhile and reminded us that we are a family.

After dinner, many families participated in a game of Family Feud hosted by the Kiel's from MI. It was great to see MSUD children and young adults and their siblings from all over the country interacting and enjoying themselves. Parents that were watching the activity couldn't resist the fun and also played the game, which was projected on a large screen in the hotel ballroom.

Saturday morning began with a wonderful buffet breakfast provided by the hotel. Those with MSUD had an opportunity to try new low protein breakfast items. Following breakfast, Sandy vanCalcar, RD from the Waisman Center, Madison, Wisconsin updated us on the nutritional management of MSUD. Her presentation was very informative and comprehensive. Brad Therrell, PhD from TX then spoke on newborn screening. We were all excited to learn that every state in the US is now screening for MSUD. We were fortunate to have Carmencita Padilla, MD from the Philippines participate in the symposium also. She shared the struggles that families in her country face each day when caring for their MSUD children. We added a new activity to the symposium this year. Just before lunch, the group played MSUD Jeopardy, which proved to be a fun, educational game. Following lunch, five teens and young adults with MSUD agreed (after some coaxing) to answer questions from the audience. Josh Drummond (CO), Lindsey Miller (PA), Ryan Merrill (UT), Lois Newswanger (PA), and Ruth Fox (WI) offered insight into how they have dealt with various MSUD issues. This is always a helpful session, especially for parents of young children. The event concluded with Delecta Li, age 10 with Classic MSUD from TN, singing "I'd Like To Teach The World to Sing". It was a wonderful weekend full of learning, sharing and fun.

Special thanks to United Services Foundation for their generous donation, which allowed many MSUD families to attend the symposium.

Plan to join us in Chicago, IL for
Symposium 2010!
Look for details in the next newsletter.



MSUD – What Happens After You Grow Up?

Symposium 08

Kim L McBride, MD
Nationwide Children's Hospital
Columbus, OH

Adolescence is a time when one's personal identity becomes solidified. Relationships outside of the family are established, and independence from parents is achieved. Teens with chronic diseases face the additional challenges of learning to manage their disease and becoming responsible for adhering to medical treatment. "Risky" behaviors common to this age group are especially risky for these kids. As one transitions from pediatric to adult care, the dynamics between the patient and the health care provider change. Communication becomes more important as the patient becomes part of the decision-making process. The challenge here is to provide high quality, coordinated, uninterrupted health care which is patient centered, age and developmentally appropriate, flexible, responsive, and comprehensive. The patient who develops skills in communication, decision-making, assertiveness, self-care, and self-advocacy will have an enhanced sense of control and interdependence in health care.

Transition will be different for an individual who is cognitively impaired. In this case, the parents may want the child to live with them longer, or may need to make a decision regarding residential vs. assisted vs. independent living. Sexual health issues are also a concern. The disabled have sexual desires, yet are at a higher risk for assault.

Once teens reach adulthood, they will be challenged to find, get and pay for health needs. Healthcare providers and institutions may avoid people with delays. The adult with a metabolic disease faces adult concerns, including the need to work, obtain medical insurance, and deal with finances and relationships. In addition, they require continued medical care. There is an absence of medical literature on the long term outcome in teens & adults with MSUD. It is known that elevated leucine levels affect the brain & nerves, so many of the problems seen in older individuals with MSUD involve the brain. In general, individuals with MSUD achieve lower levels of education, employment, and independent living. Those with MSUD suffer from higher rates of mental health problems including anxiety, depression, and possibly disordered thoughts. These conditions respond very well to counseling,

therapy and medications. Of course, a change in behavior may be the first sign of rising leucine levels. Acute decompensation happens less often, but it may be more difficult to treat.

It is important to remember that MSUD treatment only started in the 1960's, and that we are now seeing the first group of treated MSUD individuals to reach adulthood. With newborn screening resulting in earlier diagnosis, and improvements in treatment, outcomes will improve as well.

As the individual with MSUD approaches adulthood, they should work up to transition by starting to take charge of their own care. This includes tracking and planning the diet, mixing formula, and setting up a schedule to drink it and take medications. Older teens should make their own appointments and learn all they can about their disease. Give answers and ask questions.

Part of the transition includes finding a good adult physician. It will also be helpful to have an experienced social worker. It is certainly important to be a self-advocate. Adult Metabolic Clinics are rare as most metabolic specialists are trained in pediatrics. For this reason, many adults with MSUD will still be followed at a Metabolic Clinic in a children's hospital, in addition to having a primary care doctor who is trained to care for adults.

Adults with MSUD can, and have, had children. The chance of having a child with MSUD is very low (~1/850) unless the partner is also a carrier. A woman with MSUD who becomes pregnant will require careful monitoring with weekly leucine levels and periodic adjustments to the diet as leucine requirements change. It is unknown if high leucine levels cause birth defects.

Care for MSUD must be ongoing, with continued serum amino acid levels monthly. Diet restrictions are still required, and formula must be taken daily. Adults, like kids, need to have an emergency letter and an illness plan. With lifelong treatment, the outcome is expected to be good. ■

Dr. McBride cares for most of the adults in the Metabolic Clinic at Nationwide Children's Hospital, where the oldest MSUD patient is 49 years old. He previously worked as a family physician (1988 – 97), and received further training in Pediatrics, Clinical Genetics & Clinical Biochemical Genetics.

Jordan Bulcher, age 18, Classic MSUD graduated this past June from Olentangy Liberty High School in Powell, Ohio. He was active in the high school marching band and tennis team. Jordan will be attending Ohio State University this fall and majoring in molecular genetics. He'll be living on campus and rooming with a friend from high school. His parents Sandy and Dave are proud of his accomplishments and are looking forward to this new phase of life for him. They admit to struggling more than Jordan with this transition. Congratulations, Jordan!



Hi! My name is **Delecta Li**. I am 11 years old. I have a sister and she is 12 years old. She doesn't have MSUD. My mom and dad both came from China I'm half American and half Chinese. I was born in Paris,TN, and moved to Milan,TN. When I was born, I was a good baby. Then I didn't eat or drink and I cried a lot so my mom and dad took me to the hospital two times to find out what my problem was. I was in the hospital for three weeks. After three weeks the doctor told my mom that I have maple syrup urine disease. In that time my mom had never heard about that. She didn't know what to do so the doctor and the nutritionist taught my mom how to take care of



my diet. I had a hard time to get used to my formula. Often I drink 3 times a day. The formula I drink is Ketonex 2. 135g mix with 20oz. water. Every day I take 3 tablets of vitamin B-1. When I was baby, I was always sick. So we had to go to the hospital back and forth and I was late to learn how to talk or walk. I went to speech therapy to learn how to talk. When I was old enough to go to school, I went to Milan Elementary School. My mom talked to the cafeteria manager about my problem and the cafeteria started working on it. I went there from preschool through 6th grade. The low protein food there tastes good. In school, I am very good at spelling. It is always an "A". In math, I have problems so I have to really study hard. The other subjects are ok for now. During the school year, I also play gymnastics and violin. When I started gymnastics I wasn't sick very much. My health got better overtime.

This year I went to the 2008 Symposium in Ohio. I met a girl with a liver transplant and she can eat anything she likes. My goal is to be just like that. I learned that if I eat food that isn't good for me, my urine will smell really strong. Thank you for reading my autobiography! God bless you!! ■



Keeping in Touch

There is more than meets the eye when these 3 kids get together! Not only are they all about a year apart in age, they are also approximately 1 year apart in transplant!

Each summer Zac, Yasmeen and Sydney love to hang out and just be kids! Ever since they were toddlers the families try to get the kids together each year for a visit. The moms share stories, cook low protein foods (used to), or just sit and hang out where the kids can be themselves with each other.

Now as they have gotten older, their diets have changed but their love for one another hasn't. The only difference this year was they were able to enjoy hotdogs, hamburgers, corn on the cob and an endless amount of potato chips! They had fun swimming, playing on the trampoline holding the many kittens and rabbits and playing baseball with their siblings. They even put on a rock concert with Zac's instruments. You would think that they see each other all the time; they just pick up where they left off and get along great!

We are looking forward to next year!
Shawn Kelly



(Dr. Berry cont. from page 1)

For MSUD, metabolic profiling delineates the biochemical perturbations via plasma amino acid quantitation, guides in the establishment of the therapeutic plan (i.e. diet to allow for adequate endogenous protein synthesis), and allows for determination of the adequacy of treatment. As an example of the biochemical individuality which occurs with MSUD, even patients who are homozygous for the severe Mennonite branched-chain 2-keto dehydrogenase (BCKAD) E1_{sub} unit gene mutation may not manifest identical or normalized plasma amino acid profiles by simply ingesting biological protein and a medical food. Personalized nutrition is necessary to further modify amino acid intake using supplements, such as isoleucine and/or valine to maintain adequate intracellular amino acid pool sizes.

Infections cause levels of branched-chain amino acids to rise, potentially causing an acute metabolic decompensation. With a reduction in appetite, an inadequate intake of calories and other amino acids may affect hormone levels and promote breakdown of the body's own protein into amino acids, including leucine, isoleucine and valine, while depressing net protein production.

In the acute setting of metabolic decomposition, all of the above apply, but other concepts take center stage. The acute treatment in MSUD remains primarily nutritional, whether the cause is infection, fasting, accidents, or stressful events. If possible, MSUD formula is taken by mouth or feeding tube. If this is not tolerated, amino acids may be delivered parentally (by vein). Intravenous fluids with glucose and sodium bicarbonate may also be required. When the phenotype is unknown, thiamine is administered. Intracranial pressure may need to be monitored and, on rare occasions, hemodialysis may be required. In this stressed, catabolic state, cells become insulin resistant. Whole body nitrogen economy and protein turnover is affected, and there is an ever present specter of life-threatening brain edema. A judicious use of insulin is required, along with modified amino acid-containing total parenteral nutrition (TPN) solutions devoid of BCAA's, intravenous solutions of isoleucine and valine, intravenous 10-25% glucose and sodium bicarbonate, meticulous attention to salt and water

balance, fluid restriction measures, mannitol rescue, and, lastly, the employment of hemodialysis.

The conversion of branched-chain amino acids into protein requires: 1) adequate amounts in the body of all amino acids; 2) adequate amount of energy provided by dietary calories; 3) proper levels of hormones, i.e. insulin. A major therapeutic dilemma is created when patients have persistent emesis or no appetite and will not or cannot tolerate the use of a nasogastric (NG) tube for feeding purposes. The solution to this dilemma is to utilize intravenous modified parenteral nutrition. Guidelines include the 1) administration of sufficient amount of calories as glucose and/or intralipid and amino acids to satisfy minimal daily growth requirement which is dependent on age; 2) administration of amino acids in amounts sufficient to permit protein synthesis based on age dependent dietary requirements; 3) correction of insulin deficiency or resistance via continuous insulin infusion with blood glucose monitoring; and 4) use of $\geq 1.5\times$ maintenance fluids only if brain edema is unlikely or absent.

In summary, we may use modern biochemical and nutritional principles in conjunction with metabolic profiling to ensure normal growth and development in a patient with MSUD diagnosed in the newborn period. However, the risk of a serious adverse event involving brain edema, is very real despite our ability to effectively treat the patient with nutritional therapy or, even, hemodialysis. Every time a patient with MSUD undergoes acute metabolic decompensation, there is the risk of permanent brain damage or even death.

Liver transplantation, in essence, will "cure" MSUD and eliminate episodes of acute metabolic decompensation while allowing the patient to enjoy normal protein intake. The risk from surgery and its later complications and problems with life-long immunosuppressive medication are becoming less and less, I believe the risk for acute brain damage and death now outweighs the former, and therefore my recommendation is to refer patients with classic severe MSUD for liver transplantation in the first year of life. ■

References

- The Metabolic & Molecular Bases of Inherited Disease, Scriver CR, Beaudet AL, Valle D, Sly WS, Eighth Edition, 2001. (Also see The Online Metabolic & Molecular Bases of Inherited Disease)
- Inborn Metabolic Diseases, Fernandes J, Saudubray J-M, van den Berghe G, Walter JH, eds., Springer-Verlag, Inc., 4th Edition, 2006.
- Morton DH, Strauss KA, Robinson DL, Puffenberger EG, Kelley RI. Diagnosis and Treatment of Maple Syrup Disease: A Study of 36 Patients. *Pediatr.* 109(6): 999-1008, 2002.
- Kaplan P, Mazur A, Field M, Berlin JA, Berry GT, Heidenreich R, Yudkoff M, Segal S. Intellectual outcome in children with maple syrup

Symposium 08

What's New in Newborn Screening?

Brad Therrell, PhD

Professor, Department of Pediatrics,
University of Texas Health Science Center
Director, National Newborn Screening
and Genetics Resource Center

As Director of the National Newborn Screening and Genetics Resource Center in Austin, Texas, Dr. Brad Therrell was uniquely qualified to update MSUD families at our June Symposium on the state of newborn screening (NBS) in 2008. In a recent publication, Dr. Therrell and colleagues state that the goal of newborn screening “is to identify and assist all ‘at risk’ newborns and their families so that early diagnosis and intervention can occur.” (Pediatric Health 2008) Screening itself is not considered diagnostic. Rather, it detects at risk newborns who must then receive follow up so that diagnosis and the appropriate clinical management can occur in a timely fashion. Education, screening, follow-up, diagnosis, management/treatment, and evaluation are all part of the NBS system.

The United States does not have a national NBS program. Rather, it is up to the individual states to design and regulate their own NBS programs. States vary in their use of public vs. private laboratories. There are about 35 laboratories providing NBS laboratory services. NBS policy guidance is provided by professional organizations including the American Academy of Pediatrics and the Clinical Standards Laboratory Institute. All states use the services of the NBS Quality Assurance Program at the CDC for proficiency testing to ensure screening laboratory accuracy.

Most NBS programs have a group of advisors who serve as consultants and advocates. These advisory groups often consist of medical personnel along with ethicists, consumer advocates, and others. Interested parents should contact their state NBS program and volunteer to serve on the advisory group.

The National Newborn Screening and Genetics Resource Center (NNSGRC) was established by the US Department of Health and Human Services, Health Resources and Services Administration (HRSA), Maternal and Child Health Bureau (MCHB) in 1999. In a related project at about the same time, HRSA/MCHB funded the American Academy of Pediatrics (AAP) to review NBS in the US and make recommendations for improvement. This project resulted in a published ‘blueprint’ for the nation regarding NBS. As a result of this report, HRSA/MCHB contracted with the American College of Medical Genetics (ACMG) to develop a model system for evaluating conditions for possible inclusion in NBS, and to recommend a core panel of NBS conditions that should be required universally in all states. The result was a recommendation for a uniform panel including 29 core conditions and 25 secondary targets (conditions that would be identified incidental to screening for the core conditions). The core conditions include organic acid disorders, fatty acid oxidation disorders, amino acid disorders (including MSUD), hemoglobin disorders, cystic fibrosis, and hearing screening. The Secretary of Health and Human Services’ Advisory Committee dealing with NBS accepted the recommendations and forwarded them to the Secretary. States now have updated national guidance for their NBS programs. In response, all states have expanded their required screening panels, with most including the core 29 conditions. We are thrilled to announce that as of July 1 all states in the US screen for MSUD!

Research continues into other diseases including the lysosomal storage diseases. There is a concern about screening for conditions with limited treatment options, although parents have noted that screening would alleviate the need for continued medical testing in search of an elusive diagnosis. In addition, such screening could help inform future pregnancies.

Most states require screening via dried blood spots (DBS) within 24-48 hours of birth. Some states have required the collection of a second specimen to catch cases missed in the initial screening. Some cases of endocrine disorders like congenital hypothyroidism have been detected on second screening, but cases of metabolic diseases detected in this way have been rare. The facility which submits the specimen usually receives the results and has a responsibility to assist in notifying the newborn’s primary care physician. Information about positive screening findings are also reported to primary care physicians by the state NBS follow-up coordinator, usually by telephone (depending on the level of concern dictated by the screening result).

(NewbornScreening cont. on page 8)



What About Me? Supporting Siblings

Symposium 08

Karen McHugh, BS, CCLS Child Life Specialist
Nationwide Children's Hospital,
Columbus, Ohio

Karen McHugh noted that siblings of children with chronic diseases commonly feel guilt, fear, anxiety, shame, jealousy, resentment, social isolation, sadness, and anger. Siblings feel stressed over the loss of parental attention, normalcy, and a sense that parents may be indulging in the ill child. Emotions may be expressed through a variety of regressive behaviors, involving eating, sleeping, and bodily functions such as bed-wetting, fighting, and defiance, problems at school, depression, clinging, and sleep disturbances as the child attempts to gain a sense of control.

School aged children may feel a sense of responsibility for the illness of their sibling and a concern that the sibling may die. They need support to develop a clearer understanding of the illness, self-confidence, and the ability to socialize.

Supportive interventions will depend on the age. Children may benefit from expressive and sensory activities, dramatic play, play with medical equipment and supplies, music, peer to peer interactions, family activities, and bibliotherapy.

(Editor's Message cont. from page 1)

(5 months) and his mom Barbara. Six months ago Barbara had never heard of MSUD, but once her son was diagnosed she leaped in with both feet. She attended our Symposium in June, and has been busy planning a fundraiser in the Chicago area for September 26th. As though that isn't enough, she has agreed to Chair the 2010 MSUD Symposium in Chicago!



Hannah & Dylan in Chicago

So what did I learn on my summer vacation? I learned that treatment for MSUD is improving every day. For the first time, we can say that every state in the US includes MSUD in newborn screening. What an awesome accomplishment! I learned that studies are under way that will help us help our children as they navigate through school, and that, while the first generation of kids with MSUD were hampered by late diagnoses and erratic treatment, all physicians now have access to standards of care issued by the American Academy of Pediatrics. Dr. Berry's discussion of medical treatment highlighted the advances which have been made. He notes that transplantation is currently the only option to "cure" MSUD. As a family who has not chosen this route, we remain confident that treatment will continue to improve and that a relatively "normal" (whatever that is) life can be enjoyed whichever route you choose to follow.

Finally, I learned about the power of people. From the Brubachers and the Bulchers, who for years have been dedicated to helping those around the world with MSUD, to the Mudricks, with infant Dylan, who use the energy of their disbelief over a diagnosis as a means to effect change. These people are my inspiration. I hope they will be yours, too. Please volunteer. There's so much we can do together!

(Newborn Screening cont. from page 7)

Expanded screening and diagnosis has increased the need for educating pediatric health professionals about these relatively rare conditions. The ACMG has created online action/information (ACT) sheets so the health care professional can obtain additional information in a timely way and respond appropriately to a positive screening result. These ACT sheets include information on MSUD among other conditions, and a separate diagnostic algorithm is also available. Other information for professionals and consumers is available on various websites and the NNSGRC has links on their homepage (<http://genes-r-us.uthscsa.edu>).

We have moved far beyond the time when screening was referred to as "PKU screening." There is a need to educate all those who work with newborns and their parents regarding the scope of NBS and the responsibilities that attend a positive test. Electronic records will aid in the timely notification of positive screens and access to information for appropriate follow up. Geographical areas that have not had the benefit of specialized care will have increasing access to vital diagnostic information via telehealth and other electronic means. It is anticipated that access to NBS will continue to increase world-wide as more of the developing countries of the world are able to begin screening. ■

Reference: Therrel BL et al What's New in Newborn Screening? Pediatric Health 2008;2(4)



Nutrition Management of Maple Syrup Urine Disease

Sandy van Calcar

Metabolic Dietitian

University of Wisconsin-Madison

Symposium 08

Sandy provided the audience with a complete review of the nutrition management of MSUD along with her own insights based on her years of experience working with this population. The following is a summary of her talk.

MSUD Diet Basics

Metabolic formulas provide the primary source of calories, protein, vitamins and minerals for those with MSUD. Infant formulas are modeled after regular infant formulas, and provide protein (as amino acids) without leucine, isoleucine and valine. Examples include Ketonex I, BCAD I, and Analog MSUD. There are a number of formula choices for older kids, including Ketonex II, BCAD II and Acerflex. Some formulas, such as Maximaid MSUD, Maximum MSUD, MSUD Gel and Express and MSUD2 contain little or no fat. If a formula without fat is used, a source of essential fatty acids, such as walnut oil, canola, oil or flax oil may need to be given. Another formula, MSUD Amino Acid Blend, contains only protein equivalents without vitamins and minerals. It must be used in addition to other formulas, but can provide a way to increase protein without adding many calories. Finally, there are a variety of formulas used as a calorie source that contain no protein. Some of these formulas, such as Prophree contain vitamins and minerals, but others, like Polycose, do not.

If a child with MSUD drinks too little formula, then he or she may not be getting enough total protein. Too little formula can also leave a child hungry, which can result in higher levels if leucine-containing foods are eaten instead. Inadequate formula intake can mean too few nutrients, especially calcium and other minerals. To increase formula intake, different formulas, flavors, or recipes can be used, add a more nutrient-dense formula, or use a more convenient form of formula such as premeasured packets, frozen servings, coolers, or bars. Drinking too much formula, although rare, can result in an excess intake of certain nutrients that can be potentially harmful, such as excess iron and vitamin A.

Food for the MSUD Diet

When keeping track of the MSUD diet, only leucine needs to be counted since there is less valine and isoleucine than leucine in foods. The leucine content of various foods can be found in the "MSUD Food List" from Emory University or a free booklet distributed by Abbott Labs. Alternatively, leucine can be estimated from the protein content listed in "Low Protein Food List for PKU". To estimate the leucine content of a food, use these values:

Breads and cereals:	70 mg leucine/gram protein
Vegetables:	50 mg leucine/gram protein
Fruit:	40 mg leucine/gram protein
Mixed foods:	60 mg leucine/gram protein

As an example, to estimate the leucine content of Wheat Thins Multigrain crackers, use the serving size and protein content listed in the "Low Protein Food List":

1. Serving size = 2 crackers and Protein content = 0.3 grams
2. Determine the best estimate of leucine content. Since crackers are in the breads and cereals food group, multiply 0.3 grams of protein by 70 mg leucine/g protein. So: $0.3 \text{ g} \times 70 \text{ mg} = 21 \text{ mg}$ leucine in 2 crackers

The same procedure can be used to estimate leucine from food labels. BUT, protein values on labels are not exact, so the protein content listed on the label needs to be rounded up by adding 0.5 g. Then, use this protein value and multiply this by the amount of leucine/g protein.

For example, for Wheat Thins Multigrain crackers:

1. The label lists a Serving size as 15 crackers and Protein content as 2 grams
2. Round up the protein content by adding 0.5 g to the protein content on the label
 $2 \text{ g protein on label} + 0.5 \text{ g} = 2.5 \text{ g protein}$
3. Use 70 mg Leu in each gram of protein to determine the leucine content:
 $2.5 \text{ g} \times 70 \text{ mg leucine} = 175 \text{ mg}$ in 15 crackers

(NutritionManagement cont. on page 10)

Cookbooks for MSUD

In addition to several cookbooks distributed by the MSUD Parent's group, cookbooks designed for PKU can be useful:

1. Low Protein Cookery for PKU (3rd ed) (\$30)

The leucine content for all the recipes can be ordered:

Leucine content of recipes for MSUD (\$8)

2. Apples to Zucchini (\$47)

This cookbook already lists leucine values

3. What Can I Eat? A guide for adults with PKU (\$20)

To determine leucine for recipes in this book, use the same procedure used for "Low Protein Food List for PKU".

Also, using low protein products in the diet can provide an important source of calories without using up much leucine.

For example, 1 cup of regular macaroni has 456 mg leu, while 1 cup of low protein macaroni has only 30 mg.

Valine and Isoleucine

A balance of amino acids is required to form protein.

Adequate valine and isoleucine must be available to lower elevated leucine. When leucine levels are high and isoleucine and valine levels are low, additional valine and isoleucine should be added to the formula. Some physicians also recommend adding 1 to 3 grams of glutamine and 1 to 3 grams of alanine daily to provide extra amino acids for protein synthesis (Morton et al, Pediatrics, 2002).

Decreasing elevated leucine levels

If you child's leucine levels run high, but they aren't ill, then some suggestions include:

1. Increase calories by giving additional formula or adding additional fat to foods.
2. If levels remain elevated for a period of time, it is possible that the protein content of the formula is too low. Consult your dietitian about evaluating the formula content and quantity.
3. The leucine prescription may need to be decreased. This is typically not necessary unless the child's growth rate is slowing, which can happen during late infancy or late teen years.

Note that any hidden illness or infection, such as urinary tract infections (UTIs), sinus infection, menstruation, and dental problems can increase leucine levels.

Monitoring Leucine Levels

When a child with MSUD is healthy, amino acid levels are usually checked 1 to 2 times/week in infants and every 3-4 weeks in older children.

Dinitrophenylhydrazine (DNPH) is a useful urine test that can be used at home. The leucine content in blood can be estimated by evaluating the cloudiness of the urine/DNPH solution:

0	Clear	< 380 umol/l	< 5 mg/dl
+1	Slight cloudy	About 380	About 5
+2	Cloudy	400-760	5.2 - 10
+3	Opaque	> 760	> 10

Managing Illness

When the body tries to fight off an infection, this causes protein from muscle (and other places) to break down. This releases BCAA's into the blood and since BCAA's can't be metabolized by those with MSUD, leucine, valine and isoleucine level increase. Increasing calories can minimize the breakdown of protein, but this is often difficult to do if a child is unable to eat or drink. When the elevated BCAA's are converted into ketoacids, this can cause vomiting, dizziness, sleepiness, and possibly coma. All of these factors, of course, will further decrease calorie intake making the acidosis worse. Also, electrolytes (sodium, potassium, chloride) become abnormal with acidosis, causing more problems.

It is recommended that you develop a plan with your metabolic clinic before your child becomes ill:

1. Get an Emergency protocol from your clinic. This protocol should include phone numbers to page the metabolic MD in an emergency.
2. Order a Medical Alert bracelet or necklace so that if your child is in an accident and you are not there, emergency personnel will know your child's diagnosis.
3. Contact your local MD and local ER so that they know your child has MSUD. They should have a copy of the emergency protocol as well.
5. Know the process to get blood drawn and sent off for quick analysis during illness
6. Learn the process for hospitalizations in your community and your clinic's hospital so you know what to do if an admission is necessary.

A plan for illness at home

1. Start "sick diet" to increase calories and protein equivalents. This can be done by
 - a. Increasing your child's usual formula volume by 50% or concentrating the formula
 - b. Double or triple the amount of valine and isoleucine added to the formula
 - c. Remove leucine from diet, but keep calorie intake high.

This is best accomplished by taking sick day formula. If your child is hungry, provide items that contain calories, but no leucine:

- Low protein foods - add extra fat
 - Juice with Polycose (sugar), salt, baking soda
 - Gatorade with Polycose (sugar)
 - "Butter Balls" or other high calorie foods
- Caution: Pedialyte has very few calories and should not be given without additional Polycose or sugar.
- d. Tips for formula when your child is ill:
 - Small volumes, frequent drinking
 - Wake your child up at night to give formula
 - Alternate formula with juice or other beverages with calories
 - Give valine and isoleucine in juice/water if not tolerating

(NutritionManagement cont. on page 11)

(Nutrition Management cont. from page 10)

formula

- If necessary, learn to place a nasogastric (NG) tube during illness.
 - Some may need a G-tube
4. It is very important to call your clinic to let them know your child is ill:
- a. Call even if your child is “just not feeling right” but has been on sick diet for 3 days
 - b. Very positive DNPH that doesn’t improve with sick diet within 24 hours (or less)
 - c. Vomiting that doesn’t stop
 - d. Child is extra sleepy, can’t walk well

When you call the clinic, these are some things that may be recommended:

- a. Check amino acids
- b. Give Antiemetics (antivomiting medications such as Zofran)
- c. See your local MD to evaluate the illness

If a visit to the ER or hospital becomes necessary, your child may be given IV’s for calories, which can slow protein breakdown. If he or she can’t take enough formula by mouth, a NG tube or IV amino acid solutions may be necessary. The goals of a hospitalization are to reverse illness and promote anabolism (rebuilding of body tissue). Medical intervention, such as antibiotics may be required to slow/stop the illness. Nutritional management includes increasing protein equivalents so amino acids are available for protein synthesis and increasing valine and isoleucine so leucine can be utilized. If your child has an IV, this should not be discontinued until the child is able to drink 50% of his formula volume and eat sufficient calories.

MSUD Pregnancy Experience

The Biochemical Genetics Clinic at University of Wisconsin has followed 6 pregnancies for 3 women (2 classical, 1 variant). None of these women experienced any major illnesses during their pregnancies. Two of the pregnancies were notable for a slowed fetal growth in the 2nd and 3rd trimesters. Calories, BCAA’s and protein equivalents were increased during pregnancy. Leucine tolerance can increase greatly during pregnancy, especially during the 3rd trimester, because of the increased protein requirements of the developing fetus.

During pregnancy, the goals include:

1. Maintain normal BCAA levels, which decrease each trimester because of the increased blood volume of the mother.
2. Provide enough calories and protein equivalents from formula to allow for normal weight gain for the mother and growth of the baby.

Of the 6 pregnancies, all delivered at term except for one woman who delivered prematurely at 29 weeks because of poor placental function. It is not known if this early delivery was related to MSUD.

A critical time in an MSUD pregnancy is right after delivery. This is a very catabolic period (meaning the body protein is breaking down) and this can increase the leucine level and make the mother ill. For two of the 6 pregnancies, the mother developed metabolic symptoms with elevated leucine levels in the first 10 days after delivery. During these pregnancies, IV access was continued for a minimum of 24 hours after delivery and amino acids were checked 24 hours after delivery and periodically during the next 2 weeks. For post-pregnancy diet, leucine should be returned to 50% of prepregnancy intake and then changed based on blood levels.

Additional thoughts for feeding the MSUD child

Children with MSUD should be introduced to cup feeding of formula early and weaned from the bottle by 1 year of age. Present formula before food at meals, and be consistent with the feeding schedule. Do not introduce juice from a cup before starting to provide formula from the cup. Starting with juice makes it harder for a baby to learn to accept formula out of a cup.

Parents should be aware that overweight and obesity are a problem for many kids now, and prevention is key. Kids should be encouraged to maintain an active lifestyle. If a child becomes overweight, he or she should be allowed to “grow into” his weight rather than lose weight. The MSUD formula can be modified to reduce calories, but a child’s food should also be evaluated to find ways to reduce calories. ■

Dear group,

I have the grim task of letting you know that Nadya passed away on June 4th. In May, she started having swollen legs and eventually went into severe metabolic crisis. She died in her sleep at home. She was 7 years old.

Nadya was the first child diagnosed with MSUD in Russia. Anna (Nadya's mother) has put in incredible effort to make MSUD formula and information available in Russia. 3 more children were diagnosed with MSUD in Russia since Nadya, much quicker and had the formula available to them right away. Anna has translated a significant number of MSUD-related articles into Russian. The information will have a significant impact on children that are born with MSUD in Russia.

Anna is grateful to everybody in this group who has offered support, advice and help.
Irene Yusim for Anna in Russia 6-9-08



Psychological Aspects Of Adolescents With MSUD And Their Transition Into Adulthood

Symposium 08

Indira Mehta, Ph.D, Wendy Packman, J.D., Ph.D., Saman Sadatrafiel B.S.

Dietary treatment for MSUD was developed in the 1960's. At this initial stage treatment was implemented for neonates already in metabolic crises. While this treatment markedly reduced mortality, it had variable influence on morbidity and quality of life. Since then studies have attempted to identify risk factors associated with cognitive and neurological disturbances such as time of diagnosis, maintenance of dietary control and levels of leucine and its derivative levels in the blood. Collectively, studies over the last 30 years have documented improved cognitive and neurological outcomes in early-treated and optimally managed children with MSUD.

Individuals with MSUD are now generally healthy between episodes of metabolic decompensation and are expected to survive through childhood and integrate into society. There are few studies that describe the behavioral and neuropsychological sequelae in children with MSUD, and none that evaluate these issues in young adults with MSUD. Parents and caregivers have expressed concern about problems with attention, anger, violent behavior, anxiety, hyperactivity, and poor interpersonal relations in treated individuals with MSUD. With the increased autonomy and responsibilities that come with adulthood, it is important to understand whether adults with MSUD exhibit more problems negotiating adult developmental "tasks" (relationships, employment) and are at an increased risk for psychological or psychiatric disorders. This information would be helpful to healthcare and service providers as well as to family members of MSUD patients. Thus, older teen and adult patients with MSUD warrant further study.

Our study was undertaken to examine the affects of early dietary restriction of isoleucine, leucine and valine on behavior, psychological health and quality of life issues that may take place during adolescence and young adulthood in individuals with MSUD. The participants for this study were between 16-23 years of age and were recruited from the patient population followed by the Biochemical Genetic Service and Neurometabolic Clinic at the University of California at San Francisco (UCSF) and the biennial MSUD Support Group Symposium. Following informed consent, participants (patients and parents) completed a set of psychological symptom measures and a structured interview assessing their feelings about having MSUD or having a child with MSUD. The research strategy outlined above will allow for a comparison of psychometric data from individuals with MSUD and/or their parents with normative data, as well as with other inborn errors of metabolism patient samples. Understanding the psychological aspects of the transitions into adulthood for individual with MSUD will help practitioners be more sensitive to changes and transitions to adulthood and thus target interventions appropriately.

We would like to thank the families that participated in our study and plan to present our findings in a future MSUD newsletter. ■



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.

To become a member of the Spanish MSUD eGroup go to www.msud-support.org, and on the first page you will find a link to the Spanish MSUD chat group, with the Spanish and Mexican flag on it. You click there and it will take you at the bottom of the page, to the information on how to get in contact with me by writing an e-mail to monicazf@yahoo.com.



New Foods by Cambrooke Foods

Just imagine tasty Hot dogs on the grill or over a campfire...

They are now available along with Low Protein Hot Dog buns. These tasty hotdogs have only 0.5 grams of protein, 49 mg Leucine and 80 calories. The hot dog buns have 0.3 grams of protein, 21mg Leucine and 200 calories.

Have you ever wished for a peanut butter and jelly sandwich?

Now your wish can come true with Peanut Butter Spread that has only 0.1 gram of protein, 6 mg of Leucine and 130 calories per serving. This spread can also be used to make peanut butter icing, candy and cookies.

How about a tasty Blueberry Breakfast Bar to start your day out?

Just defrost in the microwave and your breakfast is all ready. These bars have only .6 grams of protein, 34 mg Leucine, and 220 calories.

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.

Please send recipes to
Food News Editor

Glenda Groff
515 W. Church Road, Ephrata, PA 17522
Ph: 717-738-4793 • ernieglenda@dejazzd.com

Peach Upside Down Cake

- 1 tablespoon butter
- 2 T tablespoons brown sugar
- 1 medium peach 150g
- 1 tablespoon blueberries
- 1/2 cup MixQuik
- 5 tablespoons water
- 1 tablespoon oil
- 1 tablespoon sugar

Spread butter on the bottom of a small single serving baking dish, sprinkle with 1 tablespoon brown sugar. Arrange peach slices and blueberries over sugar mixture. Sprinkle with remaining brown sugar. Mix water, MixQuik, oil and white sugar together.

Spoon over the peaches and blueberries, bake at 350 degrees for 20-25 minutes or until toothpick inserted in center comes out clean. Invert onto a flat plate and let the baking dish cover the cake so the sugar mixture from the bottom can soak into the cake. Serve warm with whipped topping.

	Leucine	Protein	Calories
Per recipe	98 mg	3.17 g	630
Per serving	49 mg	1.6 g	315



NEW!

COMPLEX[®] Essential
ISOLEUCINE FREE
 LEUCINE FREE
 VALINE FREE

MSD

*The **First & Only** MSUD Formula That Contains
FLAX & FIBER to Help Support a Healthy Heart*



HEART HEALTHY CHOICE

**Vanilla Flavored
 Drink Mix for MSUD**

More Options for More People™

Just in time for Thanksgiving!

Vegetable Croquettes

180 grams potatoes
2 1/2 tablespoons butter
188 grams zucchini, shredded
188 grams carrots, shredded
125 grams celery, chopped
100 grams onions, chopped
2 packets Washington's
Seasoning
low protein bread crumbs
6 tablespoons wheatstarch
3/4 teaspoon baking powder
6 tablespoons water
1/4 teaspoon salt

Cook all vegetables separately, drain well and cool. Mash potatoes adding butter but no other liquid. Combine vegetables and seasoning. Shape into balls and freeze for several hours. Combine wheatstarch, baking powder, water, salt, and sugar to make a batter. Remove balls from freezer and dip into batter and roll into low pro bread crumbs. Deep fry in hot oil until brown. Drain on paper towels. Freeze. To reheat bake at 350 degrees for 15-20 minutes. 20 croquettes

	Leucine	Protein	Calories
Per serving	17 mg	.33 g	73

Apple Salad

5 apples, peeled and diced
1- 27 oz can crushed pineapples
2 cups miniature marshmallows
2 cups Cool Whip

If apples are tart, sprinkle 1/2 cup sugar over them. Drain pineapples and mix with remaining ingredients. Chill. 10 servings

	Leucine	Protein	Calories
Per Recipe	570 mg	9 g	1780
Per serving	57 mg	.9 g	178

Note: after removing portion for low protein diet add 1/2 cup nuts to family portion if desired.

Spicy Candied Yams

16 ounce can yams or sweet potatoes
3 tablespoons brown sugar
2 tablespoons, cut into pieces
1/2 cup Rich's Coffee Rich
1/2 teaspoon cinnamon
1/4 teaspoon ginger
1/4 teaspoon cloves

Mash yams or sweet potatoes in a 1 quart casserole dish. Stir in brown sugar, butter, Coffee Rich and spices. Top with marshmallows. Bake at 350 degrees for 15 - 20 minutes, or bake at 70% power in microwave for 5 minutes. 3 3/4 cup serving

	Leucine	Protein	Calories
Per Recipe	432 mg	6.4 g	950
Per serving	144 mg	2.1 g	316

Introducing Camino Pro: Cambrooke Foods' new line of formulas for MSUD

As you can see (See page 19), at Cambrooke foods we show our commitment to the health and wellbeing of our customers through the care we take in producing the highest quality low protein foods. However, low protein foods are only part of the story when it comes to treating MSUD. No matter how good the foods are, we recognize they don't make drinking formula any easier. Therefore, we have taken our commitment to improving the MSUD diet to the next level with the development of the Camino Pro Modular Medical Food System.

Camino Pro offers those with MSUD a new way to take "formula," eliminating the bad taste, medicine-like packaging, and inconvenience of traditional MSUD formulas. With Camino Pro, you can take your "formula" in a variety of ways, so you never get bored. This revolutionary new system includes 2 drink flavors (Pina Colada and Fruit Punch), 2 Sorbet Stix flavors (Lemon Ice and Very Berry -like ice pops), and 3 salad dressings/sauces (1000 Island, Balsamic Vinaigrette, and Enchilada Sauce). Mix and match the products depending on your tastes, schedule, and mood. You'll get the amino acids you need to maintain metabolic control along with vitamins and minerals that are essential for your health.

For more information on The Camino Pro System, see our web-site at www.cambrookefoods.com/products/medical_foods/ or call us toll free today at 1-866-4-LOW-PRO.

It's time to move to the next generation of MSUD medical foods!

Swimming by Hannah Dolins

I love to swim.



When I was about 3 years old, my brother was on the Riptides White Plains swim team. My sister and I loved going to practices and meets and watching him swim. I had learned how to swim when I was 2, and I was great at it. So, when I was 4 or 5 years old, I joined the team along with my sister. And then there were 3 Dolins children on the team. Through the years we swam the best we could at meets, pushing ourselves hard at practice. Our coaches were great and encouraged us as much and as best they could. They helped us progress with each practice. When I was in 3rd grade Daniel stopped going, and then Jessica. I was the last to take a break after my 4th grade season. I came back last year again, with Jessica, to return to my team, my family. I have been with it ever since.

There is something that nobody really understands about me. It's the way I feel when I hit the water. I don't notice anything else around me, just me and my thoughts. I don't even realize how hard I'm pushing. Last year at the end-of-year ceremony, Arwa, one of the oldest members of the team, said something that I'll never forget. She said that being on the swim team isn't about swimming the fastest, or pushing yourself until you puke. It's about the friends that you make, and the songs that you sing while you're swimming. It's about the daydreams you can formulate at meets, and the fun that you have. It's about working hard to make yourself the best person you can be. This is why we love being on the team. This is why we're here.

When I'm in the water, I just swim to the beat of the songs in my head. I block everyone out and let my mind wander. I can sort out the hardest problems with ease. I feel like nothing can touch me when I'm in the water swimming. I feel like I'm in Heaven on Earth. The feeling when you know that everyone is cheering you on in meets is amazing. I try and push really hard, because I want to be great, and I want to be good and fast. One time this year I dropped about 10 seconds in 50 yards freestyle. It's great to have your friends and team and coaches complimenting you and telling you what a great job you did.

I was so happy and so was my coach Victor, but not because we probably won the meet. Victor has known me since I was 5 years old, and he was the coach who was always cheering me on and encouraging me to do my best and helping me get better in practice. I'm really grateful that he is still there and that he has supported me so much. Without him, I wouldn't have been able to be the "great" swimmer that I am today.

The bottom line is that swimming is my life. I don't think that I have ever been more comfortable and intense, and committed to a sport before. If I were just to stop swimming one day and never go back, well, I don't know what I would do because I can't even picture myself in that situation. My dad told me last year that I would never make it to even the Junior Olympics, because my muscles aren't as strong as everyone else's and so I can't do what they can do. That really upset me, because I had my heart set on going to the Olympics, but I decided that I'm going to prove my dad wrong.

It's the most wonderful feeling in the world when you swim, and I wouldn't trade it for the world. ■

The following humorous excerpt was written for the Australian Metabolic Dietary Disorders Association by parent Rachael Sharman and is reprinted here with permission.

You Know You Have a Child with an Inborn Error of Protein Metabolism When:

- You think the five food groups consist of:
1) Fruit 2) vegetables 3) rice milk 4) loprofin 5) vege chips;
- You suspect that people who don't realize bread contains protein have inferior intelligence;
- "No, it's not an allergy" becomes a daily mantra; "Yes, it's sort of like diabetes" comes in at a close second;
- You have a panic attack when your child says she needs to be sick;
- You can pronounce isovaleric academia correctly and you're game enough to have a crack at beta-methylcrotonylglycinuria;
- You don't understand why anyone wouldn't have at least 3 sets of kitchen scales;
- The top of your fridge resembles a pharmacy;
- Your 5-year-old frequently asks complete strangers - "has this got too much protein?"



Neil, multi-tasking!

Busy on the Hill: Our Efforts to Obtain National Coverage for Medical Foods

Symposium 08

Neil Buist, MD

Susan Winter, MD

Kathleen Huntington, MS, RD

This story begins about 12 years ago when Sheryl Kelsh, a parent of a child diagnosed with PKU, Kathleen Huntington and I spearheaded an effort to get a comprehensive bill in Oregon that would require all insurance issued in the State to cover medical foods. We exchanged ideas on the text of the bill over a holiday break, and eventually saw a bill passed that encompassed all Medical Foods for ALL disorders for all ages! Due to a sunset clause in the Oregon legislation, our group had to go back to the legislature in 2003 and will again have to do so in 2009 to renew the medical foods law.

A number of parent groups and clinicians have requested copies of the Oregon bill as a reference for wordsmithing their own state initiatives. Kathleen has sent out packets with samples of written testimony and summaries that are required for submission to the legislative committees in both the House and Senate before they are recommended to the floor for a vote. To date, these different legislative efforts have produced mixed results. About 35 other States have passed legislation, but many of those have restrictions on the dollar amount, products, ages or diagnoses covered.

By participating in the collaborative effort with parents and supporters and gaining an appreciation of the process required, we have expanded our efforts in getting Medical Foods properly covered, although not without some resistance from some of our own colleagues. Much of the difficulty has focused on what actually constitutes a Medical Food. The Oregon definition is precisely that of the FDA: any product that is specifically manufactured to be used under the supervision of a physician and designed to treat a medical condition.

Over a year ago, Kathleen and Neil Buist started to try and get some interest through the local offices of the NW congressional members to get a national bill introduced that would address the inequities and the state by state discrepancies. We prepared documents that showed, in great detail, what the problems were/are with the insurance coverage for Medical Foods. With great hope we canvassed several of these members and received assurances that this sounds like a real issue and that it should be relatively easy to address. FAT CHANCE!!!!!!

These efforts got us nowhere and there we would have stuck if it hadn't been for my daughter, Dr Alison Buist, who served as Senator Gordon Smith's health policy expert for 5 years – (she is now with the Children's Defense Council). We asked for her help and got more than we asked for! All of our documents were shredded into tatters by Alison, and rebuilt according to some occult formats that only DC seems to be able to generate!

Our work suddenly took on a brighter aspect when we realized that there was a bill before Congress titled "Newborn Screening Saves Lives (NBSL)", sponsored by Senator Dodd of Connecticut. What a great format for our message this should have been. However, on reading the actual text of the bill, we were dismayed to realize that the issues of TREATMENT were not addressed at all in this bill. We therefore decided to approach the problem by trying to get the terms "Long-Term Follow-up" and "Treatment" included in the NBSL act with equal emphasis as the actual NBS.

In October 07, Susan Winter and Neil Buist met in DC with the health policy experts in 14 Senate offices and were thrilled to bits when the consensus was that our points were valid and even more pleased when nearly all of the textual changes that we asked for were actually incorporated into the final text of the Bill. It was signed by the president some 2 months ago. While this is thrilling, at this time NO funding has been approved for any of the provisions in the Bill. We then started to ask whether it was possible to get support for a new Bill that would spell out the essential nature of Medical Foods and how they are comparable to any other forms of treatment. We currently have tentative interest from 2-3 Senate offices and will be working with them to see what can be done.

In the mean time there has been another development, We have been asked by the FDA to help and see if there is any way that the FDA/NIH could weigh in on the issue to clarify the nebulous status of Medical Foods, and if possible clear the way to getting some kind of national consensus for treatment coverage. This effort is in the initial stages, but we are hoping that it may be the best way forward. We will keep the MSUD community informed if and when there are any developments. ■



Anna's Make-A-Wish Trip

Anna's Make-A-Wish weekend, was absolutely amazing! I don't think anything could have made her any happier.

We flew into Los Angeles on Friday, August 22nd. Make-A-Wish arranged for a limo to pick us up to take us to the airport. Since we were flying out of Lansing, we stayed at my parents the night before. The limo was there early – 4:45 a.m.! The flight out was nice. We took the shuttle out to the Avis rental car area, and picked up the car Make-A-Wish had rented for us. We got a bit lost trying to find the freeway. The freeway... wow.... I'm definitely a small town girl at heart, because there is no way I could ever get used to that traffic!

We stayed at the Sportsmen's Lodge Hotel. It was nice, with beautiful landscaping. Of course, Anna noticed the Olympic sized outdoor pool right away! We did go swimming later that day. For me, sitting poolside, soaking up the California sunshine... yeah, that I could get used to! We were so exhausted that night. We never did get used to the 3 hour time difference. We were ready for bed at 8 p.m. California time, and I was wide away at 4 in the morning.

Saturday was WISH DAY! We were told to meet in the lobby at 10:30. There were 8 other wish families who we met and visited with. They were from all over the United States – Oregon, Florida and Texas to name a few I remember. Plus, there was a family who had come all the way over from Holland! The kids were all nervous and excited, so I think it helped them all to have others to talk to.

A Make a Wish representative met us there, and we all loaded onto a shuttle bus. We headed out to the El Capitan Theater in Hollywood. It's located right on the Hollywood Walk of Fame, across from the Kodak Theater where the Oscars are held. We were there to attend the "High School Musical 3 Ultimate Pep Rally". Two representatives from Disney, Elaine and Sal, escorted us into the theater. There were costumes on display from HSM 3. We all grabbed complimentary concessions, and were able to take our seats before the rest of the crowd.

The pep rally consisted of viewing both of the previous HSM movies on the big screen (surprisingly, Lance survived) with giveaways and surprises in between. The event was hosted by Manny, one of the DJ's on Ryan Seacrest's morning show on KIIS-FM. He gave away prizes by drawing bingo balls with numbers corresponding to the seat numbers, and also by throwing out t-shirts. The girl next to Anna caught 2 shirts, and gave one to Anna. It has Zac on the front, so of course Anna was thrilled!



Even though I've watched the HSM movies at least a hundred times with Anna, it was fun watching them on a movie screen with a theater full of people. Everyone sang along with the songs (even Lance, much to Anna's amusement and embarrassment) and screamed whenever Zac was onscreen, particularly when he was shirtless.

Besides the giveaways, when we had breaks before and after the movies, there were special guest. First three young actors who will be in HSM came out. Next were four original cast members: the actors who play Zeke (the basketball player who bakes), Jason (the sort of dense basketball player), Martha (the brainy girl who secretly loves hip-hop) and Kelsi (the composer/pianist).

Although everyone was thrilled with both of these surprises, of course, they saved the best for last. After the second movie, the previous cast members came joined by Kenny Ortega (the director), Ashley Tisdale, and of course, Zac. Wow..... I think my ears are still ringing from all of the screaming!!!! There's just something about seeing someone in person after seeing him or her on TV or the big screen – for some reason, they always look smaller in person. Ashley Tisdale is tiny! I swear, I don't think she's much bigger than Anna. And she's beautiful, even more so than in the movie. Actually, all of the girls are. And Zac – yes, he is even more handsome in person than on the screen. And yes, his eyes are really that blue. But he was shorter and thinner than I thought he would be too.

The look of excitement and happiness on Anna's face... We were shown a clip from the upcoming movie that hasn't been released before, a dance number about Prom. It was cute. After the event, Elaine and Sal ushered all of us wish families down to a little room downstairs set up with tables and an ice cream stand. We sat at a table with

(Anna's Make-A-Wish cont. page 18)



Jordan Groff was treated to a shopping trip by the Make-A-Wish Foundation at Cabela's World Foremost Outfitters. Jordan, who loves to go hunting and fishing, was able to supply himself with hunting clothes, a spotting scope, and plenty more.



(Anna's Make-A-Wish cont. from page 17)

the family from Texas, Crystal, Early and Peytlee. About 5 minutes later, Zac came in. That's when Anna's perpetual grin started.

Zac spent time at each table, talking to each wish child, posing for pictures, and signing autographs. Ours was the second table he came to. Anna posed for a picture with him right away, and wouldn't you know it, she hugged him right away. For being nervous to meet her idol, she wasn't very shy! She also did great talking to him, she didn't freak out at all. When he was signing her poster, he told her it was cool, and asked her where she got it. She matter off factly told him, "In Mt. Pleasant. At Meijers." When he was moving on to the next table, Lance and I thanked him, and he shook our hands and asked our names. Before he moved on, though, Anna did get another hug! I was very impressed with Zac. Before we met him, I was hoping that none of Anna's illusions of him would be shattered. Zac Efron isn't Troy Bolton. And there was no knowing how polite or gracious he would be. But he was very kind and friendly. I have an all new respect for him.

Shortly after Zac had started meeting with the kids, the movie's director, Kenny Ortega, came in and met with all of the families, too. He was also very friendly. And meeting him impressed me – he's choreographed some big movies, from Xanadu to Dirty Dancing. He asked Anna where she was from, and when he found out we were from Michigan, he got excited and called a young man over. His name was Tyler, but his nickname from Kenny was "Mich". It turned out he was from Walled Lake, Michigan, and was an actor in High School Musical 3. Before Zac left the meet and greet, Anna got yet another hug from him! Then, to add even more excitement to the experience – we're going to be in People Magazine!! People is going to do a special issue on HSM 3, and part of that will be a story of Zac meeting with the Make-A-Wish kids. A writer interviewed

us, and they took pictures. We'll be sure to let everyone know when the issue is going to come out!

On the shuttle ride back to the hotel, Anna complained of a stomach ache. I think it was just the release of all of the nerves she had been feeling all day.

On Sunday, we headed out to Universal Studios. We caught the shuttle bus with one of the other wish families – Diana, and her aunt and mom. They are from Miami. We saw some of the shows and did the studio tour with them. Make a Wish had not only paid for our ticket, but for the "Front of the Line" passes, which allowed us to get right to the front of the line for any ride. We felt pretty important! The studio tour was cool – we saw the outside of the soundstage that CSI is filmed at. There were trailers outside. I took a picture – who knows what actor would have been using it... Laurence Fishburne, maybe? We also saw the set of the plane crash from War of the Worlds, and the exterior set for Wisteria Lane on Desperate Housewives. Anna rode on the studio tour with Diana, which she loved. She wanted to be her adopted sister! Our favorite ride, though, was the Simpsons ride. It's a motion simulator ride, and you feel like you're right in the middle of a cartoon! We were there from 9 a.m. to 6 p.m., so it was a tiring day, but a lot of fun. After we got back to the hotel and ate dinner, Lance and Anna went down for one more swim. I, unfortunately, began the long task of packing to go home. Why is it that it is much easier to pack to go somewhere, but more difficult to pack to come home??

We are so incredibly thankful to Make A Wish. I honestly do not know how to convey in words how thankful we are, that we were given this opportunity. For a weekend, Anna didn't have to think about having MSUD, or being viewed as "different". For a weekend, she was able to feel like a VIP. I know that this is an experience that she will never forget, and it will bring joy to her heart for many years to come. ■

Beyond Low Protein: Improving the Nutritional Quality of the MSUD Diet

By Erica Lesperance Stelten, RD and Anne Kozek, RD

Many of you may recall the early days of dietary treatment for MSUD, when you had to get by with very limited resources. Low protein food choices included not-so-tasty pasta, dry bread in a can, and baking mixes that even seasoned chefs had a hard time turning into something appetizing. Formula choices were even more limited, with unflavored powder in a can often the only option. As it became evident that the diet was incredibly difficult to follow with so few allowed foods, some companies stepped up to offer more variety by producing low protein foods. The primary goal was to make foods with only the lowest protein ingredients to allow for consumption of more food without taking in more protein than the body could handle. While these foods filled a need at the time, they tended to be quite high in sugar and fat, and devoid of most nutrients, making them empty calories.

Our Commitment

At Cambrooke Foods, we feel an obligation to provide the metabolic community with more than just empty calories. We believe that metabolic diets should not only achieve the goal of maintaining good metabolic control, but they should also promote good health and prevent disease. This is why Cambrooke Foods is dedicated to creating foods that contain high quality, nutritious ingredients. To better understand how our foods will benefit your health in ways low protein foods never before have, see below.

Soluble Fiber

What it is? The indigestible component of plant-based foods. As it passes through the digestive tract, it cleanses the system and provides a number of health benefits.

Why it's important? Reduces risk of heart disease by lowering LDL or "bad" cholesterol. Regulates blood sugar.

Why it's hard to get from the typical MSUD diet? Sources are mostly restricted foods, such as legumes, nuts, barley, flaxseed, and oats.

How much you need? 25-30 grams per day (total fiber – soluble and insoluble).

Cambrooke Foods products that contain this healthy ingredient: All bread products, blueberry corn toasties, scones, toaster topz, southwestern biscuits, asian dumplings, ravioli, veggie meatballs, corny dogs, brooklyn dogs, tweekz, camburbers, medley meals, BBQ calzones, pizzas, peanut butter, santa fe biscuits.

How much do the Cambrooke Foods products contain? 2-5 grams fiber per serving, depending on the product.

DHA (docosahexanoic acid)

What it is? A type of Omega-3 fatty acid that our bodies need but cannot make adequately, so we have to get it through our foods.

Why it's important? Primary building block of the brain and retina of the eye. Associated with optimal memory function, hand-to-eye coordination, and maintaining a healthy mental state.

Why it's hard to get from the typical MSUD diet? Sources are mostly restricted foods, such as red meats, animal organs, eggs, and fish.

How much you need? No official recommendation, but suggest minimum of 160 mg per day.

Cambrooke Foods products that contain this healthy ingredient: Asian dumplings, veggie meatballs, cheese-filled veggie meatballs, peanut butter, corny dogs, brooklyn dogs.

How much do the Cambrooke Foods products contain? 32 mg per serving.

Phytochemicals

What it is? Plant chemicals that protect plants from disease, injuries, insects, drought, excessive heat, ultraviolet rays, and poisons or pollutants in the air or soil. They form part of the plant's immune system.

Why it's important? Linked to prevention of some cancers. May slow the aging process and development of age-related disease. Improve immunity.

Why it's hard to get from the typical MSUD diet? It shouldn't be, but most people don't eat enough fruits and vegetables!

How much you need? Many! No specific recommendation as there are thousands of different phytonutrient classes.

Cambrooke Foods products that contain this healthy ingredient: All products that contain real vegetable ingredients, such as: Asian dumplings, veggie meatballs, cheese-filled veggie meatballs, brooklyn dogs, corny dogs, camburbers, medley meals, BBQ calzones, pizzas, santa fe biscuit.

How much do the Cambrooke Foods products contain? Can't be quantified, but they are there!

Calcium

What it is? The most abundant mineral in the human body. 99% of it is stored in the bones and teeth.

Why it's important? Necessary to build strong bones and prevent osteoporosis. May reduce risk of high blood pressure and high cholesterol levels. May reduce risk of colon cancer. Vital for muscle contraction.

Why it's hard to get from the typical MSUD diet? Sources are mostly restricted, such as dairy products and dark leafy green vegetables. Most calcium in the MSUD diet is from formula.

How much you need? Age 1-3: 500 mg/day. Age 4-8: 800 mg/day. Age 9-18: 1300 mg/day. Age 19-50: 1000 mg/day. Age 50+: 1200 mg/day

Cambrooke Foods products that contain this healthy ingredient: All breads, blueberry corn toasties, scones, southwestern biscuits, toaster topz, santa fe biscuits, medley meals, BBQ calzones, pizzas.

How much do the Cambrooke Foods products contain? 6-15% of recommended intake of calcium per serving, depending on the product. ■

Organizational Contacts

Contact for general information:
Kay Larsen, Easton, Pennsylvania
 610-252-8572 • kaysfo@aol.com

Contact for organizational information
 & printed material:
Sandy Bulcher, *Director* • Powell, Ohio
 740-548-4475 • dbulcher@aol.com

Contact to be added to the mailing list or
 make a donation:
Dave Bulcher, *Treasurer* • Powell, Ohio
 740-548-4475 • djbulcher@aol.com

Contact for legislative & educational issues
Marcia Hubbard, *Corresponding Sec.*
 Cedar Hill, Missouri
 636-274-2021
 wingman90gl1500@juno.com

Contact for inquiries in Spanish
Monica Falconer, *Spanish Contact*
 Anchorage, Alaska
 907-770-6065 (Alaska time zone)
 monicazf@yahoo.com

Newsletter Editor
Karen R. Dolins, EdD, RD
 33 Valley Road, Scarsdale, New York 10583
 914-723-5458 • krdhed@aol.com

Medical Advisors

Holmes Morton, M.D.
 Clinic for Special Children, Inc.
 535 Bunker Hill Road
 P.O. Box 128, Strasburg, PA 17579
 717-687-9407 / Fax: 717-687-9237
 Clinic Website:
 www.clinicforspecialchildren.org

Vivian E. Shih, MD
 Massachusetts General Hospital
 Amino Acid Disorder Lab
 149 Thirteenth St., Boston, MA 02129
 617-726-3884 / Fax: 617-726-5739

Susan C. Winter, MD
 Dir., Med. Genetics/Metabolism
 Valley Children's Hospital
 9300 Valley Children's Place
 Madera, CA 93638
 559-353-6400 / Fax: 559-353-7213
 winter2571@aol.com

Neil R. M. Buist, MD
 Professor Emeritus
 Pediatrics and Medical Genetics
 Oregon Health & Science University
 buistnrm@aol.com

**Nutritional Advisors**

Rani Singh, Ph.D., R.D.
 Emory University
 Div. of Medical Genetics
 2040 Ridgewood Dr.
 Atlanta, GA 30322
 404-727-2866 / Fax: 404-727-5783
 rsingh@genetics.emory.edu

Sandy van Calcar, M.S., RD
 University of Wisconsin
 Biochemical Genetics Clinic
 1500 Highland Ave.
 Madison, WI 53705
 608-263-5981
 vancalcar@waisman.wisc.edu

MSUD Family
 Support Group Board
 Wayne Brubacher, *President*
 (Indiana)
 Ivan Martin, *Vice President*
 (Pennsylvania)
 Dave Bulcher, *Treasurer*
 (Ohio)
 Anne Fredericks, *Secretary*
 (Pennsylvania)
 Denise Pinskey
 (Michigan)
 Tony Kohl
 (Arizona)

Nutricia offers a variety of low protein foods and snacks for protein-restricted diets...

...available to purchase at MySpecialDiet.com

- An interactive, online resource for the metabolic community.
- Offers information, news, recipes and a community forum.
- Features an online metabolic shop with an extensive line of low protein foods, integral to a low protein diet.

NUTRICIA
 Metabolic Nutrition

US: (800) 365-7354
 Canada: (877) 636-2283
www.MySpecialDiet.com

MySpecialDiet.com

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

NUTRICIA
 North America
 formerly SHS North America

This publication is supported by a grant from Nutricia North America