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MSUD NEWSLETTER

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MSUD SYMPOSIUM 2016

Sandy Bulcher, VP MSUD Family Support Group and Symposium Coordinator

MSUD Symposium 2016 was held at the Embassy Suites Brier Creek in Raleigh, NC on June 23- 25. The 195 people that attended the three day event were from 21 of the 50 United States and 8 countries. Attendees included 47 individuals with MSUD ranging in age from 7 months to 47 years and 5 people that had undergone liver transplantations.

A reception was held Thursday evening. During that time, families enjoyed renewing old friendships and meeting new families. Friday was a day filled with interesting and inspiring speeches. Dr. Holmes Morton shared his perspectives on the progress that has been made in treatment from 1986-2016. Dr. Brendan Lee discussed the results of the Sodium Phenylbutyrate study that he and his colleagues conducted at Baylor. Dr. Robert Steiner of Acer Therapeutics described the company's plans to follow up on Dr. Lee's work and continue study of Sodium Phenylbutyrate as a treatment for MSUD. Dr Xinlin Hou from Beijing, China shared the challenges that clinicians face in diagnosing and treating children with MSUD in China.

Herb Foster, MSUD parent from Massachusetts, presented a check for $248,000 in honor of his son Scott C. Foster to the MSUD Family Support Group board . These funds will be used to further MSUD research. We are grateful to Herb for all of his fundraising efforts and this very generous donation.

The afternoon began with Dr. Holmes Morton who

(*Symposium* cont. on page 2)

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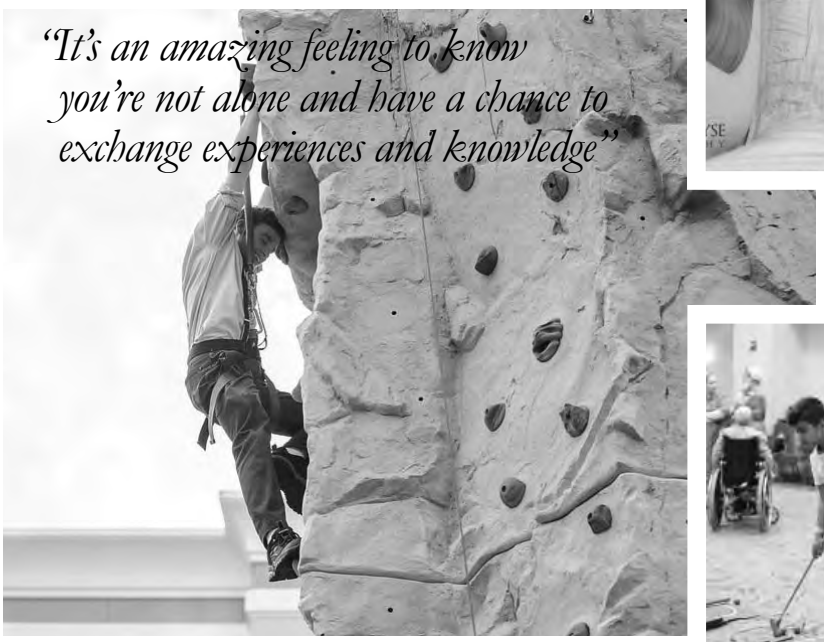
discussed his concerns regarding diet, crisis and metabolic imbalances post liver transplant. Next, the fifteen families attending the symposium for the first time introduced themselves. We were very fortunate to have Dr. James Wilson from the Orphan Disease Center in attendance.He presented next on promising research in the areas of gene replacement and genome editing. The young adult panel followed and included presentations by Seth Webb, Nikolai Rudd, Amanda Alsada, Elan Geffen, and Trey Black. This was a special time, as MSUD parents had the opportunity to ask questions of the young adults and learn from their experiences. A group photo of those with MSUD was taken prior to dismissing for dinner.

That evening, many symposium attendees enjoyed social time together. There was entertainment for all including a climbing wall, obstacle course, inflatable slide, corn hole games, and both regular and low protein ice cream. Everyone had a great time interacting and getting to know each other better.

Saturday morning began with Dr. Peter McGuire's interesting presentation on his ongoing study at the National Institutes of Health (NIH) on metabolism, infection and immunity in patients with inborn errors of metabolism . Following Dr. McGuire, Julie McClure, a metabolic dietitian from UNC Chapel Hill, presented practical information about how to manage the MSUD diet without spending a lot on specialty low protein items. The symposium concluded with MSUD Board President, Ivan Martin, sharing his vision for the support group and introducing new board members Herb Foster, Ed Fischler and Jennifer Saunders who will replace Anne Fredericks and Amy Jones. Thanks Anne and Amy for your years of service to the board and the MSUD community.

Special thanks to United Service Foundation for their generous donation which supported the travel expenses for some of the attendees. Thank you to our other sponsors, vendors and donors. Watch for the date and location of MSUD Symposium 2018 in upcoming newsletters and on our website.

Check out the MSUD website www.msud-support.org to view some of the symposium presentations in both video and PDF format. §



"It's an amazing feeling to know you're not alone and have a chance to exchange experiences and knowledge"

"It's very nice that our kids have an opportunity to meet others with MSUD and form friendships"

MSUD FAMILY SUPPORT GROUP MEMBER SURVEY

By Karen Dolins

The MSUD Family Support Group was formed in 1982 in response to a need among affected families for support and information. It is dedicated to:

- Providing opportunities for support and personal contact for those with MSUD and their families
- Distributing information and raising public awareness of MSUD
- Strengthening the liaison between families and professionals
- Supporting newborn screening programs (now a reality in every state in the US)
- Supporting research for treatment of MSUD

Much has changed in the 34 years since we first organized. More infants are being identified in the neonatal period and treatment is more standardized. Many of us connect through social media rather than through a newsletter.

To get your input on how this changing environment might affect your needs, we conducted a member survey electronically. The aim of the survey was to help us prioritize our initiatives.

We received responses from 116 individuals. Here are the results:

How important do you feel each of the following areas is for the MSUD Family Support Group to be involved in?

	Less important	Important	Very Important
Identification of metabolic specialists	**5.26%** 6	**35.96%** 41	**58.77%** 67
Research for treatment/cure	**0.88%** 1	**8.77%** 10	**90.35%** 103
Public health policy advocacy	**11.61%** 13	**31.25%** 35	**57.14%** 64
Connecting families affected by MSUD	**1.75%** 2	**22.81%** 26	**75.44%** 86
Raising public awareness of MSUD	**6.19%** 7	**27.43%** 31	**66.37%** 75

(*Survey* cont. on page 5)

How well do you feel the support group is meeting your needs in each of these areas

	Poor	Fair	Good	Excellent
Connecting families affected by MSUD	**1.74%** 2	**9.57%** 11	**46.96%** 54	**41.74%** 48
Communicating with members	**3.48%** 4	**10.43%** 12	**47.83%** 55	**38.26%** 44
Providing updates in care	**0.00%** 0	**16.67%** 19	**50.00%** 57	**33.33%** 38
Advocating for issues relating to rare diseases	**1.75%** 2	**19.30%** 22	**54.39%** 62	**24.56%** 28
Supporting research to develop treatment strategies/cure for MSUD	**2.61%** 3	**12.17%** 14	**51.30%** 59	**33.91%** 39
Improving public awareness of MSUD	**3.54%** 4	**33.63%** 38	**42.48%** 48	**20.35%** 23

Do you read the newsletter and how helpful do you find it?
89% responded that they read the newsletter and 94% reported that they find it helpful or very helpful.

If you/your child has been transplanted, do you still find value in belonging to the support group?
64 individuals responded to this question. Of those, 61% responded that they do find value in continuing to be a part of the support group.

Have you attended a symposium? If not, why?
55% Yes
45% No

Of those who have not attended a symposium, 56% reported expense and 33% reported time constraints. Many commented on the difficulties of travel, especially from other countries.

We asked for suggestions for how we can strengthen our organization and received 36 responses.
The board is carefully reviewing all of these responses.

Remember, we need an active membership to be able to follow through on important initiatives. If you feel that something is important, please consider volunteering to help. §

A Message from the Editor

By Karen Dolins

Here it is! Our one and only newsletter of 2016!

As you know, the Board decided to try producing an annual newsletter this year rather than a biannual one. This is in large part due to our ability to regularly communicate with the community through our website and 900+ member Facebook group. However, we do recognize that not everyone is connected electronically and feel that it is important to continue to produce a print newsletter at least once a year.

This year's newsletter is full of information about our symposium which took place this June in Raleigh, North Carolina. It was wonderful to reconnect with everyone in our MSUD family and to meet new family members. Some of our speakers agreed to be video-taped, and you can hear their presentations in their entirety on our website at www.msud-support.org. Summaries are also available in this newsletter.

In this issue you will also find the results of our first ever electronic member survey. Thanks to all of you who responded!

This past April I represented our support group at the Genetic and Metabolic Dietitians International Symposium (GMDI) in Scottsdale, Arizona and describe my experiences in this issue. While at the symposium, I met dietitian Keiko Ueda and learned of the new Metabolic Diet App Suite which she helped develop. I hope you'll read the article she wrote describing this new tool and how it can help you manage the MSUD diet.

We were sorry to hear of the untimely death of Jennie Verbeek. Her parents beautifully describe her life and passions in this issue.

Amber Raye tells us of her accomplishments, and we are introduced to new Board members Herb Foster, Ed Fischler and Jennifer Saunders.

Some of you have asked for an expanded Diet Wise section. Unfortunately, our previous contributor has stepped down and no one has taken her place. Anyone interested? If you are, please contact me via email (see below).

Until the position is filled, please feel free to take a look at the Diet Wise section on our website where you will find a link to NBS-Connect (https://nbs.patientcrossroads.org) and their library of low protein recipes.

Finally, on a personal note, our daughter Hannah (classic MSUD - 22 years old) continues to do well. If you were at the symposium, I'm sure you met her! She had a wonderful time socializing with all of you.

Daniel, Jessica and Hannah Dolins visit Paris

Taking a break from her ongoing college studies, she had a great time this spring when our family visited Paris and Wales where her brother and sister were in school. Of course we made contact with local metabolic clinics before we left but fortunately we didn't need to visit!

This newsletter is not the work of 1 person. Thanks to each of you who contributed, and a special thanks to Sandy Bulcher and Yossi Dworcan for their expert proofreading; and to Eddy Wang for his expertise in designing the layout producing the finished product. If you enjoy writing and would like to contribute to this newsletter or even help edit it, please contact me at karen.dolins@yahoo.com. §

From the Chairman's desk

By Ivan Martin

As I reflect on the Symposium from this past June in Raleigh, North Carolina, I feel the time and effort needed to go was well worth it. The subjects presented were very informative and gave those of us whose lives revolve around MSUD new hope and strength to continue striving for better clinical care and ultimately a cure for MSUD. I believe we are on the verge of some very exciting discoveries in the medical field including gene therapy.

Thanks to the United Services Foundation, we were again able to assist ten families from all over the world with travel expenses to attend the symposium. Some of the families traveling from foreign countries have very poor clinical care facilities;for those families, the Symposium is a very informative and a much needed educational opportunity for them.

The MSUD Board of Directors has reorganized by accepting the resignation of two of its long time members and has added three new members. Thank you Amy Jones and Anne Fredericks for your years of service. We welcome Jennifer Saunders, Edward Fishler and Herbert Foster as our new board members.

During the symposium, Herb Foster presented a giant check to the MSUD Family Support Group from the Scott Foster Foundation. These funds are earmarked specifically for medical research. Our hope is that through this generous contribution, we can move closer to our goal of finding a cure for MSUD.

Thank you again to all those who were able to attend the symposium. §

News & Announcements

Bausell in Israel

Dietitian Heather Bausell, of Lurie Children's Hospital in Chicago, Illinois, traveled to Israel where she visited the metabolic clinic at Schneider Children's Medical Center in Tel Aviv. While there, she met Dr. Avraham Zeharia (above), staff, and some patients. §

Katelyn DeGaetano Age 32 Classic MSUD

My husband, Peter, and I are excited to announce the birth of our second child. On March 10, 2016, I gave birth to a little boy named Pietro Scott via C-Section at Brigham and Women's Hospital in Boston, Massachusetts. Baby Petey is a very happy and healthy little boy. Throughout my pregnancy, I was monitored closely, but did not have any complications due to MSUD. Baby Petey was delivered early for other reasons, but we both did very well. I guess he could not wait to meet his family including sister Kayla (3)! §

INTRODUCING THE **NEW BOARD MEMBERS** OF THE MSUD FAMILY SUPPORT GROUP

Edward (Ed) Fischler is the father of David Fischler, who has classic MSUD. Ed is married to Lynn Fischler and together they have three children - David (age 27), his twin brother Michael, and a daughter, Jennifer, age 29.

Ed is a native of Virginia. After serving in the Army in the early 1970's and later graduating from the College of William and Mary in Virginia, he moved to Atlanta, Georgia. There he met and married his wife, Lynn. Their children were all born and raised in the Atlanta area. In 2014, after concluding his 37-year career with Southern Company, he and Lynn moved to Windermere, Florida, where they presently reside. Ed plays golf and volunteers for roles in local charity golf tournaments and with Lynn, they both enjoy traveling. Ed, Lynn, and David attended their first MSUD Family Support Group symposium in Philadelphia in 2014. At that conference, David spoke about managing MSUD while in school and college, his experiences playing on his soccer team with his brother and high school teammates, and marching in his high school band and later with the Georgia Tech marching band.

Like others affected by MSUD, David, Lynn and Ed anxiously await a cure. In the meantime, the MSUD family support group is an important - if not the most important source of information (and comfort) about how to manage illnesses, diets, as well as tips, suggestions, and advice from other parents.

Herb Foster is the father of Scott C. Foster (deceased) and Katelyn DeGaetano, both with classic MSUD. "I have been given the honor and privilege to be made a member of the MSUD family support group Board of Directors. I hope to be active with the research committee and help all members of the support group with any fundraising ideas they would be interested in undertaking on behalf of our new research fund."

Jennifer Saunders is a 48 year with classic MSUD. She lives in Phoenix, Arizona. She has attended most of the MSUD Family Support Group symposia. §

The Metabolic Diet App Suite: MSUD Diet Tracking on the Go!

By Keiko Ueda MPH RD, Gloria Ho, Roderick Houben, Jeffrey Joa, Alette Giezen RD, Barbara Cheng RD, Clara van Karnebeek, MD, PhD, FCCMG

Lifelong medical nutrition therapy (MNT) is a key part of medical therapy for many inherited amino- and organic acidopathies. But there is a lack of metabolic disorder specific diet intake tracking and meal planning resources. Many individuals living with metabolic disorders experience difficulties following their daily metabolic diet goals.

The Metabolic Diet App Suite is a free, web-based metabolic diet tracking tool developed to help people living with metabolic disorders track and plan their daily diets. It was created with input from biochemical geneticists, metabolic dietitians, metabolic patient caregivers, and application developers for use on both mobile devices and desktop computers (www.metabolicdietapp.org).

General disorder information is provided for fifteen individual metabolic disorders including: 'MSUD' (Maple Syrup Urine Disease), 'PKU' (Phenylketonuria), and a general protein tracker 'PROT'. Food nutrient content is based on the MetabolicPro™ program food database, compiled by the Genetic Metabolic Dietitians International (GMDI) Technology committee. The MSUD Diet App offers functions such as: secured personal user login/password, user instructions, goal setting, daily leucine (milligrams) and protein (grams) diet intake tracking, food content checks, adding foods and homemade recipes, exportable daily food diary log (to update your doctor and dietitian), and developer feedback. Individuals can start by going to the website and creating a personal account and diet profile, no downloading required.

MSUD METABOLIC DIET

www.metabolicdietapp.org

The website provides a user guide: 'How it works?' with answers to frequently asked questions (FAQs) such as how to recover forgotten password and usernames and contacting the developers. Initial pilot tester feedback was positive and suggestions used to further improve the App suite.

This project was funded by the British Columbia Children's Hospital Foundation (Treatable Intellectual Disability Endeavor in British Columbia, www.tidebc.org, 1st Collaborative Area of Innovation) and the Rare Diseases Foundation (Vancouver, Canada).

The Metabolic Diet App Suite is intended as a patient and caregiver support tool. They do not replace the GMDI Metabolic Pro™ diet analysis program or health care professional advice. We hope that this tool helps ease the daily challenge of metabolic diet tracking and meal planning for individuals living with protein or amino- or organic acid restricted metabolic nutrition therapies.

Ho G, Ueda, K, Houben RFA, Joa J, Giezen A, Cheng, B, van Karnebeek CDM. Metabolic Diet App Suite for inborn errors of amino acid metabolism. Molecular Genetics and Metabolism. 2016 Mar;117(3):322-7. §

Gene Replacement and Genome Editing
in the Treatment of Liver Metabolic Diseases - Are We There Yet?

By James M. Wilson, MD, PhD
Director, Orphan Disease Center
Director, Gene Therapy Program
Medicine and Pediatrics Perelman School of Medicine University of Pennsylvania
Summarized by Sarah Dworcan

Dr. Wilson has worked in the field of gene therapy for the past 35 years. He readily accepted the invitation to join our conference as he felt it was time to share the progress which has been made in the development of gene therapy, and to consider the possibility that this may become an effective treatment for patients with MSUD. Are we there yet? In Dr. Wilson's words: "I leave it up to you to decide whether we're just around the corner or not". His goal was to update us on the progress of gene therapy across other disorders, specifically as it relates to treating liver metabolic diseases.

What is Gene Therapy?

All metabolic processes in our bodies start with a gene in the DNA. The gene makes messenger RNA (mRNA) and the mRNA makes a protein. DNA remains present for the life of the cell, but mRNA is only present when a protein needs to be made.

In MSUD, there is a defect in one or more of the 4 genes which produce the branched-chain alpha-keto acid dehydrogenase complex (BCKDC) needed to metabolise the branched-chain amino acids. Due to the defective gene, the correct mRNA is not produced and therefore the incorrect protein is produced. This causes a block in metabolism and leads to the accumulation of toxic leucine.

The treatment for most metabolic diseases is to try to manipulate the accumulation of those metabolites with drugs or diet. This requires ongoing treatment and diet regimens that last a lifetime. Gene therapy aims to treat the disease at its root, in the gene. Dr. Wilson states that in MSUD "the normal version of the gene is the ultimate drug". If we can insert a new form of this gene into the cell, to reside there permanently, we essentially correct the genetic defect.

What is the Challenge of Gene Therapy?

The normal version of the gene for BCKDC is known; the problem is getting it into the right cell. At the turn of this century it was discovered that packaging the DNA in the right way enables it to overcome the barrier of the cell membrane. Once through the cell membrane, the package containing DNA with the normal gene takes up residence in the cell, produces the correct protein, overcomes the block and diminishes the accumulation of leucine.

mRNA Therapy

Research has demonstrated that through mRNA therapy, the correct protein to overcome the metabolic defect can be produced. However, mRNA therapy is not permanent, and eventually the treatment is reversed.

Host Viruses

So how do we package DNA to get it into cells? Viruses. Viruses don't have DNA of their own, making them a great package to shuttle the correct gene. Jean Bennett, an ophthalmologist, was the first to successfully use gene therapy through a vector system, improving retina sight for blind people with a specific genetic mutation. Despite the noted success, a good vector for the liver is still not available.

(*Gene* cont. on page 11)

The problem with most host viruses is that they are too large to pass through the cell membrane. The advantage of the liver and what makes it a great organ for gene therapy over other organs is that the liver blood vessels have large pores in them. When a substance is injected into the blood it goes directly to the liver, not other organs such as the heart. With this in mind Dr. Wilson's team treated mice with Hemophilia B, a genetic disease affecting the way the blood clots. This was successful, and they have since gone on to use this treatment in humans.
This successful vector gene therapy in Hemophilia not only saves $300,000 per patient per year, more importantly, the patients do not require repeated infusions and are essentially cured of their disease. Many biopharmaceutical companies have since pursued gene therapy for Hemophilia using vectors.

Dr. Wilson says that what the researchers and the MSUD community need to look at now is whether this is a viable treatment for MSUD. The delivery vehicle, regulatory genes and treatment would be essentially the same as for hemophilia. The defective/mutated gene would be swapped for the normal version of the gene.

Dr. Wilson notes that a patient who has been exposed to the virus that is used for the vector may have an immune response, causing the body to reject the virus and the treatment. Otherwise vector gene therapy has shown to be quite safe.

Genome Editing

An alternative to gene therapy is genome editing. Experimentation on Urea Cycle Disorder, another metabolic disorder, has shown that gene therapy may work with a mature liver but may not work in an infant whose liver is still growing. Since gene therapy introduces the new gene into the cell to float in the cell, it doesn't attach to the DNA. With an infant, as each cell divides and grows it will grow more of the new normal gene but also more of the old defective gene and eventually there will be more cells with the defective gene than the normal gene.

Genome Editing is different from gene therapy in that it not only delivers the normal gene to the cell but it actually goes in and corrects the mutation, which in turn corrects the mRNA, which produces a healthy protein. This would be the best approach for a newborn liver while gene therapy would theoretically work from 3 years of age and onwards.

Targeted Genome Editing

The challenge with genome editing is that each mutation requires a different drug. Researchers needed to find a way to use genome editing without being mutation specific. With this in mind, they successfully inserted the normal version of a gene into the chromosome of test mice. Unlike gene therapy where the gene is outside the chromosome, here the gene was actually inserted into the right place of the chromosome, so that it could function independent of the specific mutation. The survival and success rate of the mice was extremely high.

Orphan Disease Center

Going forward Dr. Wilson and his colleagues are using the positive data from the Hemophilia study, changing the gene, and leveraging the experience gained to multiple diseases. They have made the appropriate vectors for MSUD and will soon begin test runs in the mouse model. Dr. Wilson is quite confident that treatment in the mouse model will be successful and that human trials will follow.

Dr. Wilson closed by mentioning how humbling it is to see the commitment of physicians and caregivers, and the support that all provide to one another. He promised to continue to try his best, be transparent and keep the MSUD community informed. In his words: "I just hope to God it succeeds!"

Dr. Wilson's research has been made possible by the University of Pennsylvania, Orphan Disease Center. §

MSUD DIET ON A DIME

By Julie McClure, RD

Specialty manufactured low protein foods have long been an important part of dietary management for MSUD and other disorders of amino acid metabolism. They can be helpful at providing appropriate calories and the leucine content of each item is readily available.

However, these products can be expensive for those who do not have insurance or state coverage for the specialty manufactured low protein foods. Fortunately in the last few years as vegan, gluten-free and even paleo diets have become more popular, it has become easier to find foods in regular and specialty grocery stores that can work for the low protein diet.

Although typically more expensive than regular grocery store items, these grocery store finds are often cheaper than specially manufactured low protein foods. You can take home your items with the same day you shop, with no shipping charges either!

Protein values are estimates from a variety of sources (howmuchphe.org, food companies, USDA database or calculations from ingredients and nutrition labels). Check labels carefully to ensure you have the correct product. Protein on the nutrition label should be consistent with amount you are counting. As a general rule of thumb you can multiply the protein grams by 60 to get an estimated mg leucine per serving. Check with your metabolic team if you have questions. Let's go on a quick tour of what you may be able to find.

Item	Brand	Location	Serving Size	Protein
Produce and Breads				
Cassava (Yuca) Root		Hispanic GS	½ cup	1.4 gm
Mountain White Bread	Canyon Bakehouse	Target	1 slice	1.3 gm
Tapioca Loaf	Ener-G	WF, EF, GC	1 slice	0.7 gm
"Seafood and Meat"				
Breaded Vegan Scallops	Sophie's Kitchen	WF, online	8 pieces	0.2 gm
Breaded Vegan Coconut Shrimp	Sophie's Kitchen	WF, online	4 pieces	0.8 gm
Jackfruit BBQ, TexMex, Curry, Chili Lime Carnitas, Original	The Jackfruit Company, Upton's	WF, EF, some WM	2.5 oz	1.3 – 1.8 gm
"Dairy"				
Coconut Milk	Silk, So Delicious	GS, WM, Target	8 oz	0.5 gm
Almond Milk	Silk, Blue Diamond	GS, WM, Target	8 oz	0.7 gm
Flax Milk	Good Karma	GS	8 oz	0 gm
Rice Milk	Dream	GS, WM, Target	8 oz	0.5 gm
Coconut Milk Yogurt	TJ, So Delicious	TJ, GS, WM, EF, WF	5 oz	0.5 gm
Almond Milk Yogurt	Dream	WF, EF	6 oz	1.4 gm
Coconut Milk Ice Cream (without added nuts or chocolate)	TJ, So Delicious, Coconut Bliss	TJ, WF, EF, GS	½ cup	0.7-1.4 gm
Almond Milk Ice Cream	So Delicious, Dream	WF, EF, GS	½ cup	~ 1.3 gm

(*Diet on a dime* cont. on page 13)

Rice Milk Ice Cream	Good Karma, Dream	WF, EF, GS	½ cup	0.4 – 1 gm
Cheese (slices, shreds or blocks)	Daiya	WF, EF, some WM, Target	1 oz (28 g)	1.4 gm
Chao slices (creamy original or herb	Field Roast	WF, EF, some WM	1 slice (20 g)	0 gm
Chao slices (tomato cayenne)	Field Roast	WF, EF	1 slice (20 g)	0.2 gm
Imitation Slices or Shreds (Mexican, America or Italian)	Sunny Acres	Dollar Tree	1 oz (28 gm)	1 gm
"All New" Cheese Blocks or Slices (American, Garden herb, Mozzarella, Provolone)	Follow Your Heart	WF, EF, some WM	1 oz (28 gm)	0 gm
Vegan Gourmet Blocks "Made with Organic" (Monterey Jack, Nacho, Mozzarella, Cheddar)	Follow Your Heart	WF, EF,	1 oz (28 gm)	1.3 gm
Shreds (Fiesta blend, Mozzarella, Cheddar)	Follow Your Heart	WF, EF	1 oz (28 gm)	0.2 gm
Imitation American Slices	Sandwich Mate or Smart Options	WM or other GS	1 slice (19 gm)	0.5 gm
Frozen Foods				
White Sandwich or Multigrain bread	Glutino	WF, EF, GS	1 slice	0.9 gm
Gluten-Free Original Bread	Rudi's	WF, EF, GS, WM	1 slice	0.5 gm
Gluten-Free Waffles	Trader Joe's	TJ	1 waffle	0.6 gm
Homestyle Waffles	Nature's Path	WF, EF, GS	1 waffle	0.5 gm
Fried Plantains	GOYA	GS	1 piece	0.3 gm
Ripe Plantains	GOYA	GS	5 pieces	0.5 gm
Broccoli or Sweet Potato Littles	Dr. Praeger's	WF, EF, GS	2 pieces	1 gm
Puffs (Taro, Carrot or Kale)	Dr. Praeger's	WF, EF, GS	7 pieces	1 gm
Salty Snacks/ Starches				
Gluten-Free Pretzels	Snyder's	WF, EF, GS, WM	1 oz	0.2 gm
Pretzels	Glutino	WF, EF, GS	1 oz	0.5 gm
Gluten-Free Pretzel Crisps	Snack Factory	GS	1 oz	0.3 gm
Original, Black Pepper, Toasted Onion	Absolutely Gluten Free	WF, EF, GS	9 crackers	0.3 gm
Flatbread- Everything	Absolutely Gluten-Free	WF, EF, GS	2 flatbreads	0.3 gm
Original or Table Crackers	Glutino	WF, EF, GS	1 oz	0.7 gm
Bagel Chips	Glutino	WF, EF, GS	1 oz (7)	1 gm
Sai-Fun Noodles	Ka-me	GS	1 cup cooked	0.1 gm
Mai-Fun Noodles	Ka-me	GS	1 cup cooked	0.7 gm
Shirataki calorie free pasta	Miracle Noodles	GS	1 cup cooked	0 gm
Cassava (Yuca) Chips	Arico	GS	1 oz	0.4 gm
Plantain Chips	Trader Joe's	TJ	1 oz	0.5 gm
Sweets				
Lemon Wafers	Glutino	WF, EF, GS	1 wafer	0 gm
Kinnitoos Chocolate or Vanilla Sandwich Cookies	Kinnikinnick Foods	WF, EF, GS	1 cookie	0.2 gm
Toaster Pastry	Glutino	WF, EF, GS	1 pastry	0.9 gm
Peanut Spread	Walden Farms	GS	1 Tbsp	0 gm
Biscoff Spread/ Cookie Butter	Lotus, Trader Joe's	GC, TJ	1 Tbsp	0.5 gm

Key: GS- Grocery Stores (which ones vary depending upon product and your region), EF- Earth Fare, WF- Whole Foods, TJ- Trader Joe's, WM- Walmart

FROM THE PROFESSIONAL JOURNALS

Heterozygote to homozygote related living donor liver transplantation in maple syrup urine disease: A case report.

Patel, N., Loveland, J., Zuckerman, M., Moshesh, P., Britz, R., & Botha, J. (2015). Pediatric transplantation, 19(3), E62-E65.

This case report describes the liver transplantation of a 2.5 year old child who received a portion of his mother's liver. An initial graft rejection was successfully managed, and the child has been able to tolerate a normal diet without protein restriction. He lost weight initially and developed a respiratory tract infection, but blood BCAA levels have remained normal. The authors suggest that relatives can be a safe source of donor livers when an unrelated liver is not available.

Acute Metabolic Crisis in Maple Syrup Urine Disease After Liver Transplantation from a Related Heterozygous Living Donor.

Al-Shamsi, A., Baker, A., Dhawan, A., & Hertecant, J. (2016).

This case report describes a metabolic crisis in a 20 month old child who had received a liver transplant 5 months earlier from a parent, who carried the defective MSUD gene. The child did well after transplant until developing a gastrointestinal infection. Blood levels of leucine, isoleucine and valine were all elevated indicating an inability to completely process branched-chain amino acids during illness, and the child experienced seizures and encephalopathy.

Individuals undergoing liver transplantation typically receive livers from non-related donors who do not carry the gene for MSUD. As both parents must carry this gene for a child to have the disease, parents have historically not been considered as donors. More recently, though, there have been a number of reports in which parents have successfully donated a part of their liver to their child with MSUD. The authors of this case report recommend continued metabolic monitoring post-transplant, particularly in the case of a parent donor and when infections occur.

Development of Carrier Testing for Common Inborn Errors of Metabolism in the Wisconsin Plain Population

Kuhl A, van Calcar S, Baker M, Seroogy CM, Rice G, Schwoerer S (2016). Genetics in Medicine Aug 11. doi: 10.1038/gim.2016.104.

A statewide outreach project was developed through the University of Wisconsin Biochemical Genetics Clinic and the Wisconsin Newborn Screening Program to identify members of the Plain population who are at risk for having children with maple syrup urine disease (MSUD) or propionic acidemia (PA). Blood spot testing kits were distributed through health care providers and information about the initiative was provided through state midwives and Plain population meetings. Eighty individuals were tested, and genetic counseling and follow up diagnostic testing was provided for those identified as at risk for having a child with PA (none were identified as at risk for having a child with MSUD).

Early identification of at-risk couples will allow for early treatment of at-risk babies during the newborn period, improving long term outcomes. §

Advocacy Update

By Jordann Coleman, MSUD Family Support Group Advocacy Chair

There are a variety of ongoing issues that face the MSUD and greater rare disease community. While we push forward to improve the lives of those with these diseases, here is a quick update on the some recent advocacy issues that are facing the rare disease community:

- The Senate Authorizes Medical Foods Coverage for the Military in National Defense Authorization Act. This bill allows for medical foods to be covered under the TRICARE health care program for Department of Defense and Military families. The bill will need to be harmonized with the House version passed a month earlier.
- The Senate delayed their vote on the Senate Innovations for Healthier Americans Initiative, also known as the Senate Cures Legislation. Senate Cures is the companion legislation to the 21st Century Cures Act, which passed the House last year. The legislation includes billions of dollars to help the rare disease community, including new funding for critical research at the National Institutes of Health (NIH) and to accelerate drug approval at the FDA, and several other provisions. Rare Disease advocacy groups will continue to push for this legislation to pass once the Senate is back in session in September.
- The State of Illinois is waiting for a bill to be signed by the governor that would establish a Rare Disease Commission that would give patients a voice in the state government. The bill would also provide educational resources for elected leaders on critical issues related to access, coverage, and the diseases themselves.
- A Rally for Medical Research Day will be held on September 21, 2016 on Capitol Hill. Over 300 organizations will participate to urge members of Congress to make funding for the National Institutes of Health (NIH) a national priority and raise awareness about the importance of continued investment in medical research. To sign up for the "Rally" reception or the "Rally Hill Day", visit www.rallyformedicalresearch.org.
- There are several patient groups in Oregon seeking to reduce the out-of-pocket drug costs for patients. The Oregon legislature recently created a Prescription Drug Cost Working Group who met for the first time this past June. The National Organization for Rare Disease (NORD) is supporting policy recommendations, which encourage the State Working Group to address cost barriers and other issues that impede patient access to medicines. If advocates are interested in learning more about the Working Group and these issues, contact Tim Boyd at tim.boyd@rareaction.org

Larger issues like the Medical Foods Equity Act and better timeliness with Newborn Screening are works in progress. If you are interested in advocating for MSUD in your state or federal governments, please contact me: coleman.jordann@gmail.com. §

Enhancing Pediatricians Awareness of MSUD Infant Health in China - What Can We Do?

By Xin-lin Hou, MD,PhD, Pediatric Department, First Hospital, Peking University, Beijing, China with Karen Dolins

The first case of MSUD in China was reported in 1987 and described a girl in Shanghai whose parents were first cousins. Prior to her birth, three of her brothers had previously died of unknown causes. The diagnosis of MSUD was made at Xin Hua hospital, Shanghai Jiao Tong University by a blood test for branched chain amino acids.

No further cases were reported for 8 years, but 46 cases have been reported since 1994 and several papers describing MSUD diagnosis, treatment and prognosis in China have been published, including one by our hospital in the European Journal of Medical Genetics in 2015. Most diagnoses have been made in the Eastern area of China around Beijing and Shanghai. There are almost no cases reported from mid and Western China, where the economy is less developed. We believe that more children have been born with MSUD but have not been diagnosed.

Clinical features of children with MSUD in China are not unique, but the time of diagnosis and treatment is always relatively late. Most patients were admitted to the hospital with neurological symptoms 7 days after birth, including poor feeding, seizures, coma and respiratory failure. Of the 40 cases of neonatal onset, 28 died, 5 had combined developmental lag and epilepsy, and 7 cases were lost to follow up. Among the 7 children who lived for more than 1 month, 1 has died, and 4 were lost to follow up. All of those diagnosed with MSUD who lived for more than one month have mental retardation and epilepsy.

Treatment for MSUD in China remains difficult as the metabolic formula is not available. Although newborn screening has identified a number of cases, the prognosis is poor. One paper reported on newborn screening at Shanghai Xinhua Hospital, which is the transfer center of inborn metabolic disorders for the whole country. They screened 410,000 newborn babies and found 3 cases of MSUD, but did not mention the prognosis. Another article screened 19,000 newborn babies, found 4 cases of MSUD, but the prognoses were all poor.

We comprehensively analyzed 33 patients diagnosed with MSUD. There were 28 cases of classical type, presenting with dystonia, sleepiness, poor response, poor feeding, seizures, retardation, and varying degrees of metabolic acidosis. Of the 33 cases, 10% of them were Vitamin B sensitive (400mg per day) and 5 cases were intermediate type (2 cases were adult, one is 29 years old, the other is 30 years old).

In 2012, one patient was diagnosed with classic MSUD at 18 days of age. He was initially diagnosed with neonatal pneumonia due to vomiting and feeding difficulties, but soon lapsed into a coma. Gene sequencing identified a BCKDHA compound heterozygous mutation with a C.740 A>G mutation on one chromosome and on another chromosome IVS6+1G was missing. The family was able to obtain the MSUD formula through contacts outside of the country. Formula was bought in the UK and shipped to Hong Kong, where it was then brought over to China. The child improved, but several weeks after diagnosis and initiation of the diet severe diarrhea and a rash appeared. We researched the literature and determined that the likely cause was a lack of isoleucine and valine. After adding some ordinary formula milk for 5 days, the diarrhea and rash were cured.

At the age of 1 year and 5 months, he experienced a bout of diarrhea and refused formula. He rapidly decompensated, became hypoglycemic and ketoacidotic, and lapsed into a coma. He was treated with hemodi-

(*China* cont. on page 17)

alysis and BCAA levels decreased to the normal range. This case is unique to us as the parents have been able to cover the cost of care including measurement of BCAA levels, which is not covered by health insurance.

The prognosis of children with MSUD in China is poor for the following reasons:

1. Lack of routine screening. Most children were identified after the appearance of clinical symptoms.
2. Delay of 2-4 weeks in obtaining laboratory results. Also, most laboratories can only assess leucine and valine levels, not isoleucine and alloisoleucine.
3. BCAA free formula, isoleucine and valine, and low protein food for MSUD patients is not available..
4. The vast majority of hospitals can not perform liver transplantation.

Our hospital has treated a total of 8 cases of MSUD since 2012. One case was diagnosed with newborn screening and 7 cases were diagnosed with selective screening upon presentation of neurological disorders. Of these, 6 cases were classic and 1 case was intermediate. In one mother's second pregnancy, we ran amniocentesis and gene analysis and determined that the fetus was heterozygous. This baby was born healthy. This is the first time we have had a successful prenatal diagnosis. We have also succeeded in performing liver transplantation for two girls, whom are still in follow-up.

We wish that in the near future, the government could provide free routine screening to every neonate, BCAA-free formula, supplementation and health insurance coverage for treatment.

Overall, for MSUD in China, from the government to the pediatrician, the understanding of MSUD is still in the initial stage. We hope that pediatrician's understanding of MSUD can be improved, early diagnosis and early treatment can be implemented, and the metabolic formula essential to survival will be made available in order to improve survival rates and reduce occurrence of neurological disability. §

Genetic & Metabolic Dietitians Int'l Meeting

April 2016

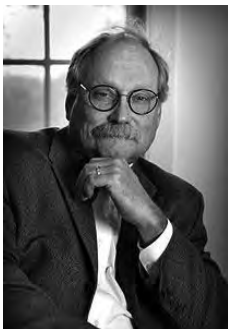
By Karen Dolins

Genetic and Metabolic Dietitians International (GMDI), the professional group to which most of the dietitians you work with belong, holds conferences every other year. This April I represented our group at their conference "Metabolic Nutrition in the 21st Century: Looking Towards the Future," held in Scottsdale, Arizona. The purpose of my attendance was to exhibit and network; to teach dietitians about our support group, learn about advances in the field and network with representatives from other support groups.

As an exhibitor, I provided attendees with our brochure, research highlights, information about our group, and information about our upcoming symposium. Most were familiar with our group but weren't aware of all we do. Others did not know about us and were happy to take information so they could refer families with MSUD to us for support. Dietitians expressed an interest in partnering with us for the development of educational materials in different languages, especially Spanish and Arabic.

In speaking with representatives of other support groups, I found that advocacy and fundraising were key focuses. While the PKU organization is unique in that they have a paid executive director, most rely on the dedication of volunteers as we do.

At this conference I learned of the Metabolic Diet App Suite, developed by dietitian Keiko Ueda and colleagues at British Columbia Children's Hospital. Please read Keiko's article in this newsletter describing the app, as I believe many of you will find it useful in dietary compliance. §



Dr. Holmes Morton MSUD 1986-2016: A Sense of Progress

Summarized by Karen Dolins

Dr. Morton provided attendees with a history of MSUD treatment in the United States. He reports that 1/2 of the children born with MSUD between 1965 and 1986 died of cerebral edema.. Now, in 2016, hospitalizations have been drastically reduced and outcomes greatly improved. It is important to monitor growth charts for evidence of sustained, normal growth.

The Clinic for Special Children in Lancaster, PA has provided accessible and affordable care to over 2200 patients with 127 different recessive disorders, including 90 Mennonite patients with classical MSUD.

One area where there has been great improvement is in our understanding of the effects of high levels of branched-chain amino acids (BCAAs) on the brain. Leucine has a strong affinity for the protein (LAT 1) which carries it into the brain, so high levels cause the brain to be flooded. At the same time, the entry of other amino acids becomes blocked, creating an imbalance which affects brain chemistry. One consequence of this is the dystonia (muscle spasms) observed when levels are high.

Patient outcome has also improved due to recognition of the importance of supplementing with valine. Previously, valine deficiency led to impaired head and brain growth. Valine has low affinity for the transporter, therefore brain levels are very low when blood leucine levels are high.

Another advance has been the use of BCAA total parenteral nutrition (TPN), which allows for successful surgery and rapid reduction of leucine levels. Dr. Morton suggests that hospitals keep premeasured and sterilized individual amino acids in storage so

(*Morton* cont. on page 19)

this option will always be available as an alternative to ordering liquid TPN, which only has a shelf life of about 1 month. In his opinion, this is much safer than dialysis. When cerebral edema is present he suggests giving mannitol, a sugar with high osmolality that is used with head injuries, as it pulls water out of brain and reduces cerebral edema. Lasix (a diuretic) and sodium chloride should also be given to remove extra fluid and prevent hyponatremia. An individual in a metabolic crisis must be made anabolic (building rather than breaking down body tissues) so they can build protein and reduce blood leucine levels.

Dr. Morton's second talk of the day addressed transplant and other treatment modalities. He works closely with the transplant team at Children's Hospital of Pittsburgh, where they have conducted 72 MSUD transplants without any fatalities or need for retransplantation. After transplant, patients are immediately able to process normal amounts of protein and maintain normal blood leucine levels. However, neurological problems including anxiety, mood disorders and sleep disturbances remain. Livers from MSUD patients can be safely transplanted into other patients as most of the enzyme to metabolize BCAAs is in muscle and other tissue.

There have been 3 reported cases of transplanted children who developed encephalopathy after an illness accompanied by dehydration. BCAA levels in these children were elevated, but dropped after IV rehydration. Dr. Morton noted that the transplanted liver can metabolize dietary protein adequately but it has a harder time when the body is in a catabolic state such as occurs with infection. This is especially difficult with dehydration. It is essential to monitor amino acid levels during infections, even after transplant.

Amino acid imbalances in the brain appear to persist after transplant. Dr. Morton believes that supplementation with specific amino acids will enable the brain to make neurotransmitters and may help with neurological function.

Stem cell transplantation as a possible therapeutic technique has received attention recently. However based on Dr. Morton's experience this will only modestly increase BCAA tolerance.

Dr. Morton closed by noting that liver transplant is a drastic intervention using a scarce resource. Follow up is intensive, requiring frequent monitoring and multiple medications. Families shouldn't give up hope for alternate therapies such as gene therapy where progress has been seen. §

Thank You

Our sincere gratitude to the following sponsors:

- Charles Hehmeyer (travel expenses)
- Children's Hospital of Pittsburgh of UPMC
- Nutricia North America
- United Service Foundation (travel expenses)
- Vitaflo USA
- Acer Therapeutics (Friday evening ice cream bar)
- Cambrooke Therapeutics (Low protein foods)

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Thanks to our exhibitors:

- Acer Therapeutics
- Cambrooke Therapeutics
- Children's Hospital of Pittsburgh of UPMC
- NBS Connect
- Nutricia North America
- Vitaflo USA

Metabolism, Infection and Immunity in Inborn Errors of Metabolism

By Peter J. McGuire, MS, MD
National Human Genome Research Institute (NHGRI)
National Institutes of Health
Summary by Karen Dolins

Dr. McGuire is conducting research at the National Institutes of Health (NIH) on the interaction between the immune system and metabolism in individuals with Inborn Errors of Metabolism (IEM). He notes that decompensation resulting from infection is more severe than decompensation due to improper diet, and believes that the way in which the immune system and IEMs impact each other is largely responsible.

Our immune system protects us against bacteria, viruses, fungi, parasites, cancer, and pollution. We have multiple lines of defense. These include our skin, tonsils, lymph nodes, and special cells called inflammatory cytokines which are soluble mediators made by immune cells that allow communication between cells. These inflammatory cytokines are responsible for the fever and loss of appetite that can accompany infection.

Infection is harmful for individuals with MSUD and other IEM as it increases energy (calorie) requirements. Breakdown of muscle protein is increased as the body tries to generate this extra energy, releasing branched-chain amino acids which can then cross over the blood brain barrier and enter the brain itself.

Dr. McGuire's research subjects have had other IEM. He is now looking to enroll individuals with MSUD so that he can study the impact of this metabolic disease on the immune system in hopes of improving the understanding and treatment for metabolic crises. If you are interested, please contact him at: ministudy@nih.gov. §

Acer Therapeutics Continues Phenylbutyrate Research

By Robert D. Steiner, MD
Chief Medical Officer, Acer Therapeutics
bsteiner@acertx.com
715-650-3030

We at Acer Therapeutics (www.acertx.com) are delighted to have the opportunity to inform the MSUD Community about our work in development of a pharmacologic therapy for MSUD. I am pleased to have been invited to discuss our work at the 18th BIENNIAL MSUD SYMPOSIUM in Raleigh, North Carolina in June. We look forward to continuing collaboration with the MSUD Family Support Group. Our success at Acer is dependent in part on our responsiveness to the MSUD patient and physician community's needs. Strong advocacy support from clinicians, patients, families, and patient advocacy groups is critical to the success of companies like ours.

I will begin with a quick introduction to our company and who we are. Acer is a specialty orphan pharmaceutical company. Our strategy is to develop therapies for the treatment of serious, ultra-rare diseases with critical unmet medical need. The medicines we are developing have some degree of established safety data from animal (pre-clinical) and human (clinical) studies undertaken previously. Typically, clinical proof-of-concept studies have already been completed prior to our involvement. This approach reduces development risk, timelines and cost.

Acer's headquarters are in Cambridge, MA. We have a very experienced management team including:

(*Acer* cont. on page 21)

- Chris Schelling, CEO, Founder and Director who has 17 years of biotech/pharma strategic and orphan drug expertise; Chris was previously at BioMarin
- Jeff Davis, Head of Corporate Development who has 25+ years of drug discovery, business and corporate development expertise; Jeff was previously at Genzyme
- Harry Palmin, Acting CFO spent 15 years as President, CEO and Board Director at Novelos Therapeutics, oversaw 10 clinical studies in oncology and infectious diseases; previously at Lehman Brothers and Morgan Stanley
- Ben Dewees, Vice President, Regulatory & Manufacturing, who has 20 years of orphan drug regulatory and manufacturing expertise at BioMarin and Biogen Idec
- Robert Steiner, MD, Chief Medical Officer, who has 20+ years orphan disease treatment and research experience including MSUD. Bob is Clinical Professor at the University of Wisconsin, having previously held positions at Marshfield Clinic and Oregon Health & Science University (OHSU)

Acer currently has two drugs in the pipeline, for three orphan diseases. ACER-002 is being developed for vascular Ehlers Danlos Syndrome (vEDS) and will not be considered further here. ACER-001 is being developed for Maple Syrup Urine Disease (MSUD) and also Urea Cycle Disorders (UCD). ACER-001 is a taste-masked, immediate-release formulation of sodium phenylbutyrate (NaPB). As some of you may know, NaPB is a pretty horrible tasting drug! So, we have spent a couple of years developing a formulation of NaPB that has very little, or no taste at all. This is important, because we want patients to take all of their medicine, and that may be difficult to do if the drug tastes bad.

NaPB was originally approved by the Food and Drug Administration (FDA) in 1996 to treat UCD. UCD are life threatening disorders of amino acid metabolism, not unlike MSUD (1). Acer is currently collaborating closely with Dr. Brendan Lee and Baylor College of Medicine, where NaPB is currently being studied for treatment of patients with Maple Syrup Urine Disease (MSUD). As the readership of this newsletter knows, MSUD is an inborn error of amino acid metabolism (1). There are currently no approved medication treatment options and dietary therapy alone is not optimal. Patients may suffer chronic neurological damage and life-threatening metabolic decompensation. Our goal at Acer is to develop ACER-001 to be used in addition to dietary therapy in treatment of MSUD. It was noted by physicians at Baylor College of Medicine (BCM) and elsewhere several years ago that NaPB reduces branched-chain amino acid levels (2). Leucine (Leu) is a branched-chain amino acid that accumulates in patients with MSUD and is responsible for complications. Lowering Leu in MSUD is beneficial. Brendan Lee and colleagues at BCM identified the novel mechanism of action of NaPB in MSUD – NaPB inhibits the branched-chain ketoacid dehydrogenase kinase that, in turn, activates the branched-chain ketoacid dehydrogenase complex – which is the enzyme deficient in MSUD – thereby potentially increasing the effectiveness of the MSUD enzyme (3). A clinical proof-of-concept study in MSUD with NaPB carried out at BCM showed statistically significant decreases in Leu (3). 5 MSUD patients received 3 days of a steady-state protein diet followed by 3 days of NaPB plus diet. The results showed a statistically significant decrease in Leu in 3 of 5 MSUD patients ($p < 0.05$ in responders). Leu levels fell approximately 30% (28-34%) in responders. Clinicians consider a 20-30% decrease in blood Leu levels as clinically meaningful.

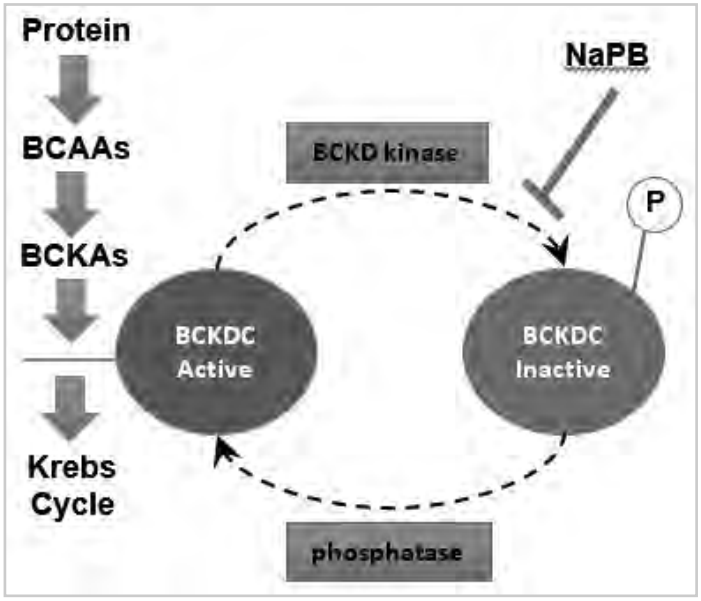


Figure showing NaPB inhibition of BKCD kinase and its downstream effects

(*Acer* cont. on page 22)

A second trial of NaPB in MSUD is underway at BCM. We at Acer are planning a Phase 2 study of ACER-001 in which we will evaluate the degree and frequency of response to ACER-001 treatment as demonstrated by reduction in blood Leu levels among MSUD patients with elevated blood Leu levels. We will evaluate the safety of ACER-001 treatment in MSUD patients, identify individuals with MSUD who respond to ACER-001 with a clinically-meaningful reduction in blood Leu, and evaluate the correlation between blood Leu levels and branched-chain α-ketoacid dehydrogenase (BCKD) gene mutations. Current enrollment plans (which are subject to change) include pediatric and adult MSUD patients with elevated Leu levels. Approximately 60 patients will be enrolled. Safety is assessed by medical history, monitoring adverse events and vital signs, physical examinations, lab tests (e.g. amino acids, chemistry, hematology, and urinalysis). Approximately 10-15 sites will be needed, chosen from clinics in around the US with experienced MSUD clinicians.

Acer Therapeutics

In summary, Acer Therapeutics (www.acertx.com) is a specialty orphan pharmaceutical company whose strategy is to develop therapies for the treatment of serious, ultra-rare diseases with critical unmet medical need. We are developing a new taste-masked, immediate release formulation of sodium phenylbutyrate (ACER-001) for treatment along with diet of MSUD. We very much look forward to partnering with the MSUD medical, scientific and patient communities in development of this new, exciting therapy.

References:

1. Rice GM, Steiner RD. Inborn Errors of Metabolism (Metabolic Disorders). *Pediatr Rev.* 37:3-17. 2016
2. Burrage LC et al. *Mol Genet Metab*. 113:131-5. 2014
3. Brunetti-Pierri N et al. *Hum Mol Genet*. 20:631-40. 2011

Amber Raye

Classic MSUD, Age 30

My name is Amber Raye and I have classic MSUD. I am currently working towards my master's degree in Forensic Psychology at Southern New Hampshire University with a current GPA of 3.8. I hope to graduate in the summer of 2017. Having MSUD, though challenging, has not by any means discouraged me from my hopes to obtain my dream of working in forensics. I obtained an opportunity to intern for a few days at a county morgue in New York beside my best friend, in order to gain a better understanding of the human body and forensics in the area of forensic medicine. After I graduate I hope to attend Philadelphia College of Osteopathic Medicine to obtain a second master's in forensic medicine.

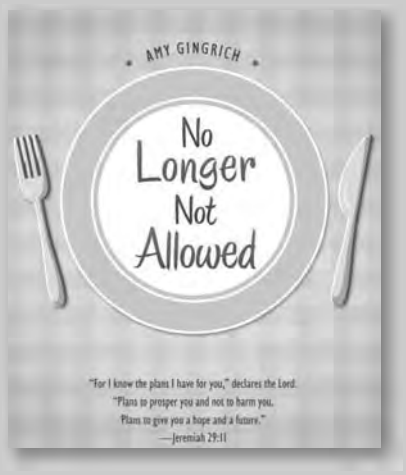
As a child with MSUD I went through extensive trauma when I was given the wrong medicine at the age of seven years old. I spent a year in the hospital with high leucine levels and fluid in my frontal lobes, which is exceptionally dangerous for anyone. During my time in the hospital I experienced horrific hallucinations which resulted in a lengthy list of phobias upon my discharge from the hospital. This proved to be disabling for a young girl at such age. So along with the struggles of MSUD and learning to cope with food restrictions, I was diagnosed with non-military related Post Traumatic Stress Syndrome (PTSD). Despite being advised to take multiple anti-anxiety medications to help curb the symptoms of PTSD however, I refused to take them. I spent the better half of 23 years overcoming my phobias and conditioning myself to utilize my experiences to better myself and make it a goal to fully understand them. It is because of my drive and creativity that I can say with a smile that 95% of my phobias are now manageable to deal with. I still struggle when triggered and still have symptoms that do create challenges, but I continue to strive to understand the mechanics of them. The most important and incredible aspect of my experience is that I managed to not sustain extensive damage to my frontal lobes during my trauma.

I have learned that as an individual with MSUD I do have limitations but also have realized that creativity to adapt and overcome makes the worse days manageable. I do not settle for impossibilities. I strive to better my health by walking everyday and eating healthy foods. I watch my diet and learned from my trauma the warning signs that my levels are escalating. I also learned to be my own advocate when it comes to seeing my specialist. I continue to educate myself and keep myself up to date by reading various journals regarding my disease and I couple that with my experiences to avoid falling into a similar experience. §

READING LIST - *No Longer Not Allowed - $15*

In ***No Longer Not Allowed***, Amy Gingrich shares her story of living with the disease and then undergoing a liver transplant at the age of twenty-one in January of 2006 at Pittsburgh Children's Hospital. She tells about the hospital stays, the ups and downs of healing, subsequent health issues related to the transplant, and recovery. Gingrich also includes recipes and journal entries from her parents who provided unconditional support.

Signed books can be purchased directly through Amy (Amy Zimmerman Gingrich, 605 South Cedar St, Lititz PA 17543. Phone # 717-341-1383) or at Amazon.com (If purchased from Amazon, she will sign it at the 2018 symposium).



Our Daughter Jennie

By Betty and Dirk Verbeek

Long before we were ready, we sadly said goodbye to our daughter Jennie Verbeek, who was involved in a serious accident on June 11, 2015. She was hit by an automobile while crossing the road on her mobility scooter.

Jennie was born November 20th, 1973. She was 7 days old when she was diagnosed with Maple Syrup Urine Disease. This made her the first diagnosed case in Ontario and even though the prognosis was poor at the time, she was able to beat the odds. They started treatment at 8 days of age, but a day later she had 5 Cardiac Arrests, which left her with mild Cerebral Palsy.

She grew up a happy girl and her dad nicknamed her Smiley because her smile was her trademark. The doctors were always delighted with her blood results and when we asked if she should have a liver transplant, they did not recommend it since she meticulously followed her prescribed diet. As soon as Jennie became a teenager, she started to keep a diary of everything she ate. If she was short on equivalents she would make it up by adding peas. Her favorite vegetables were fried potatoes and turnips. We could always tell when her levels were off because of the missing sparkle in her eyes.

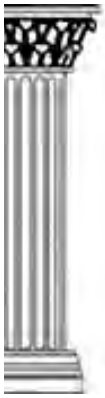
Jennie enjoyed a very active social life; she made many friends, because of her bubbly and outgoing personality and her fun-loving spirit. She never let MSUD prevent her from going places or doing things she loved.

Jennie loved to travel and had been to Tennessee on a school trip, with the family to Aruba, twice to Europe and visited many of the cities where the symposiums were held. She was so looking forward to attending this year's MSUD symposium and to visit Dollywood on the way back home. She never travelled without her doctor's 'To whom it may concern-Letter' in case of an emergency.

She enjoyed a great number of hobbies ranging from piano lessons, bowling, card making, shopping, crocheting and riding her scooter - to show her independence.

Jennie had a love for people and had the gift to gab. She truly enjoyed having a conversation with anyone who would join her. Her willingness to serve in any way possible made her a big part of our church community and our community at large.

My husband and I could never have asked for a more wonderful daughter. Everything she did made us proud. We take comfort in knowing that she is safe in the arms of Jesus and that our much beloved little girl made a true difference to others. Jennie's big wonderful smile, her unconditional love, caring and the beauty she brought into our lives will live on forever. §



Maple Syrup Urine Disease & Social Security Benefits

By Bryan Mac Murray, Outreach Specialist, Social Security Disability Help

If your child has been diagnosed with MSUD, Social Security may be able to help with the costs of treatment. The process may seem daunting, but it may help to know which benefits you would be able to apply for and how you can apply for them.

Which Social Security Program should I apply for?
The Social Security Administration (SSA) has two programs to help with disability benefits, Supplemental Security Income (SSI) and Social Security Disability Insurance (SSDI). SSDI is for adults who have been employed, so a child would not be eligible for these based on their own work record. However, if a parent is receiving disability or retirement benefits, the child may be eligible for certain SSDI benefits.
Children of a SSDI recipient who are under 18 years of age may be eligible for benefits based off of their parent's work record, regardless of whether the child has a disability or not. This is because these benefits are provided as part of the parent's benefits to cover the cost of raising a child. With children over 18, they must have been disabled before the age of 22, and they must meet the Social Security Administration's definitions of disability. Generally, these benefits would be around 50 % of the main beneficiary's (the parent) monthly benefit amount, in addition to the parent's monthly benefits.

Supplemental Security Income (SSI) is a Social Security program intended to support low-income families whose children or parents have disabling condition. There are strict financial requirements to qualify for SSI based on income, beyond the Social Security Administration's disability criteria. Generally, if a family is earning less than $1,100 a month and has less than $3,000 in resources, they may qualify for SSI benefits. However, after the age of 18, the Social Security Administration would consider the child's own income and resources when determining benefits eligibility, rather than the parent's income and resources, even if they live in the same household.

How does the Social Security Administration determine disability?
The Social Security Administration determines disability based on both medical criteria and on an assessment of a person's capacity to work. For children with MSUD, the SSA would determine their eligibility based on the medical criteria outlined in two documents: The SSA Blue Book, and the Compassionate Allowance List.

Is MSUD listed in the Social Security Blue Book?
Social Security's general Blue Book doesn't list MSUD specifically, but the illness can certainly qualify if it meets certain conditions found under Childhood Blue Book Sections 111.06- Motor Dysfunction (Due to any neurological disorder) and 112.02 A1 (Organic Mental Disorders):

- 111.06 requires persistent problems with motor function of two extremities. This must affect major daily activities and disrupt fine and gross movements, or gait of station.
- 112.02 A1 requires medical documentation showing persistence of developmental arrest, delay, or regression.

Does MSUD qualify for a Compassionate Allowance?
While it's not listed in the Blue Book, the SSA considers MSUD severe enough to be included it in their Compassionate Allowance List. This means your claim evaluation process will be expedited.

Evidence of MSUD must be provided, as per the SSA's Compassionate Allowance Guidelines for MSUD (https://secure.ssa.gov/apps10/poms.nsf/lnx/0423022445). This consists of documents showing tests and exams that led to the diagnosis of MSUD. You'll need to contact your child's physician for these documents.

(*Social Security* cont. on page 26)

Your child's diagnosis of MSUD automatically qualifies him or her as having a disabling condition. With a Compassionate Allowance, your claims process can be reduced to as little as a few weeks and you could start receiving benefits right away.

Applying for benefits based off of a parent's work history:

If you are receiving SSDI or retirement benefits, your child may be eligible for the benefits described above. To apply for these types of benefits, schedule an appointment at your local SSA office. You can also apply for these benefits online, at the SSA's website. You can find frequently asked questions, and checklists for applying for benefits as well.

Applying for SSI benefits

To apply on behalf of your child for SSI benefits, you'll need to locate your local Social Security office. You must make an appointment and apply in person. You'll need to gather special information before going to the appointment. Bring this information with you:

- Your child's date of birth, birthplace, and Social Security number.
- Contact information for your child's referring physician, or a specialist, who can help Social Security with the application and knows about your child's condition.
- Contact information, patient ID numbers, and dates of treatment for all doctors, hospitals and clinics.
- List of medicines your child takes and who prescribed them.
- Names and dates of medical tests your child has had, and who referred you.

Don't hesitate to apply for these benefits for your child, as they can help your child get the care they need to stay healthy. Social Security can help pay for care and treatments so that you can focus on your child's health.

About the Authors

Disability Benefits Help is an independent organization that is dedicated to providing information about Social Security Disability benefits and how to qualify for them with debilitating illnesses and conditions. To learn more, please visit: www.disabilitybenefitscenter.org §

DietWise

Many thanks to *Dana Angelo White* for submitting recipes!

See more at:
http://danawhitenutrition.com/recipes/

Our readers appreciate sharing in any tasty recipes you have found or developed, so please send them in!

Easy Balsamic Vinaigrette

Serves 12

Ingredients:

- ¼ cup balsamic vinegar
- ½ cup extra virgin olive oil
- Juice of ½ a lemon
- 2 teaspoons honey
- 1 clove garlic (whole, lightly smashed)
- ¼ teaspoon kosher salt
- 1/8 teaspoon freshly ground black pepper

Directions:
Combine ingredients in a jar or other airtight container. Cover, shake well and serve. Store in the refrigerator for up to one week.

Nutrition Info per serving:
Calories: 80; Total Fat: 9 grams; Saturated Fat: 1 grams; Carbohydrate: 1 grams; Protein: 0 gram; Cholesterol: 0 mg; Sodium: 25 mg; Fiber: 0 grams

URL to article:
http://danawhitenutrition.com/recipes/easy-balsamic-vinaigrette/

Sweet Potato Chips

A healthy and fast dinner side dish that everyone will love.

Serves 6

Ingredients:

- 3 large sweet potatoes, unpeeled
- Olive oil
- Sea salt

Directions:
Slice sweet potatoes very thin (a hand-held mandoline slicer works perfectly). Place on a baking sheet, brush with olive oil and season with salt. Bake for 15 to 20 minutes until crispy and golden.

Nutrition Info per serving:
Calories: 76; Total Fat: 2g; Carbohydrates: 13g; Protein: 1g; Leucine: 60 mg; Isoleucine: 40 mg; Valine: 60 mg

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