

# MSUD NEWSLETTER

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**MSUD Symposium**  
Save the Date:  
**June 28-30, 2012**  
at the Embassy Suites- Airport  
Philadelphia, Pennsylvania



## The Inborn Errors of Metabolism (IBEM) Collaborative:

Metabolic clinicians in the Region 4 Collaborative (Illinois, Indiana, Kentucky, Michigan, Minnesota, Ohio, and Wisconsin) and colleagues in six other states (New Jersey, Pennsylvania, New York, Missouri, Oklahoma, and South Dakota) recently learned that they have received a five-year grant from the National Institutes of Health to study long-term outcomes for children affected with conditions identified by newborn blood spot screening. Directed by Principal Investigators Cynthia Cameron and Susan Berry, this grant support provides exciting new opportunities for improving outcomes for children identified by newborn blood spot screening (NBS). All of these states screen newborns using tandem mass spectrometry (MS/MS) to identify rare, serious

IBEM including MSUD.

While long-term follow-up is critical for monitoring health outcomes and evaluating the effectiveness of newborn screening, standards of clinical care for screened conditions have never been subjected to evidence-based study. More information

(IBEM cont. on page 2)

*Editor's Note: Dr. Zinnanti traveled to Columbus, Ohio to meet with several board members and present his research hypothesis and methods. The MSUD Family Support Group board was impressed and excited by his presentation, and decided to support his research with a grant for \$20,000.*

## New Research for Prevention and Treatment of Brain Injury in MSUD

Dr. William Zinnanti

Child Neurology Fellow, Stanford University Hospital



It has been nearly 60 years since Dr. John Menkes first published his description of four cases of a rapidly progressive, lethal encephalopathy with a peculiar odor, eventually identified as maple syrup urine disease (MSUD). Since this disease was recognized, much progress has been made by many dedicated physicians and scientists in both our understanding of its disorder has an incidence of 1:180,000 in the general population, with a much higher frequency in certain Mennonite and other groups.

MSUD results from a deficiency of the enzyme branched-chain ketoacid dehydrogenase. This truly remarkable molecular

(Dr.Zinnanti cont. on page 3)

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# Impact of Inborn Errors of Metabolism on Admission in a Neonatal Intensive Care Unit- A Prospective Cohort Study

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## Abstract Objective

To estimate the incidence of Inborn Errors of Metabolism (IEM) in neonatal intensive care unit (NICU) using tandem mass spectrometry and to determine the impact that these disorders have on NICU resources.

## Methods

During the period of study, 957 babies were admitted to the NICU. Of these, 724 had dried blood filter-paper samples collected and analyzed for Inborn Errors of Metabolism using tandem mass spectrometry. The diagnosis was further confirmed through clinical symptoms and by gas chromatography-mass spectrometry. The results were also confirmed by clinical follow-up of all positive patients in an overall interval of 1 year. The mean observation period was 11 months per neonate.

## Results

In total, 22 cases screened positive and 8 cases of inborn errors of metabolism were detected. The incidence of IEM in the population of patients admitted to the authors' NICU was 1.1%. The most common inborn error found was methylmalonic acidemia (3 cases, 37.5%). All of the cases needed aggressive treatment and invasive mechanical ventilation. There were two cases of Tyrosinemia type 1, one case each of Maple Syrup Urine Disease, Propionic Acidemia, and Multiple Acyl-CoA dehydrogenase deficiency (MADD). Five of the eight patients required invasive mechanical ventilation. The median length of NICU stay was 3 days (1~7 days) and early therapeutic intervention was effective for four of them and other four patients (50%) died.

## Conclusions

The incidence of IEM in NICU was 1.1%, indicating an underestimation of the incidence of metabolic disorders prior to implementing screening. Most patients with IEM in the NICU required invasive mechanical ventilation and the mortality was increased due to underlying IEM.

Only 1-2 patients were diagnosed with IEM prior to the use of tandem mass spectrometry.

(IBEM cont. from page 1)

about outcomes for these disorders is essential to a better understanding of the natural history of the conditions and development of best practice models for treatment. The grant allows collection of information about the health outcomes, complications, and life progress of persons that have conditions identifiable by NBS.

This grant provides funding to continue work initiated by the Region 4 Genetics Collaborative Priority 2 Project Workgroup that began building an IBEM Information System with support from the Health Resources and Services Administration (HRSA) Maternal Child Health Bureau Genetic Services Branch. More than 300 persons have already agreed to participation in the project and have allowed collection of ongoing information about the rare IBEM affecting an individual in their family.

With the receipt of this grant, additional families who attend clinics guided by participants in this research will be invited to participate. Participation consists of allowing the site investigators and their team to abstract information about the progress and medical needs of the affected person. Information is stored in a secure, password-protected database; identifiable information about individual persons enrolled in the project is available only to the site investigator responsible for the individual enrolled. Privacy is carefully protected; information about individual participants will be examined in aggregate so that participating centers can accrue information about these rare conditions.

Over time, we hope that the data collected in this project will build the foundation for evidence-based medical practice and care for

rare disorders ascertained through NBS because they will provide data to support treatment decisions based on larger cohorts of affected children than can be seen by any individual practitioner or specialty center. With the collaboration of multiple centers over time, a dynamic, longitudinal database will have the power to provide a foundation for evidence-based medical practice and care for rare disorders ascertained through newborn screening.

Our concept is that by examining where differences in treatment plans correlate with changes in outcome we can build an evidence base for optimal treatment choices. Further, if families agree, participation in the project provides a point of contact for other research activities and a means to facilitate data collection about those related research activities. We hope to extend our efforts to collaboration with other researchers interested in new strategies for improvement in care for affected persons. We hope this research project will help both clinicians and researchers learn more about the real lives and outcomes of people who have these conditions. ■

For more information, please see <http://region4genetics.org/>

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(Dr.Zinnanti cont. from page 1)

machine shares the same super structure and components as the two other highly conserved (structure maintained through many species) keto-acid dehydrogenase family members, alpha-ketoglutarate and pyruvate dehydrogenase. Stationed within the mitochondria or "power plant" of the cell, the keto-acid dehydrogenases are intimately associated with the basic metabolic function of the cell to produce energy. In the brain, the keto-acid dehydrogenases not only produce energy but also maintain the homeostasis of glutamate, the chief excitatory neurotransmitter of the brain.

I was introduced to MSUD as a medical student at Pennsylvania State University while following one of Dr. Holmes Morton's patients who was in a coma from encephalopathic crisis for an entire month. I watched this child return to normal after a month of what appeared to be certain death from uncontrolled brain swelling. From this experience, I was moved to pursue research in MSUD. The establishment of the Clinic for Special Children in Pennsylvania by Dr. Morton has been instrumental in improving outcomes. Dr. Morton has also raised awareness of MSUD by continued collaborations with local world class research centers such as Children's Hospital of Philadelphia, Pennsylvania State University and University of Pittsburgh. These efforts have recruited additional help. Dr. Harry Paul, Dr. Kristen Skvorak and Dr. Greg Homanics developed the MSUD mouse model at the University of Pittsburgh. By studying the brains of these mice, we learned that leucine levels rose exponentially in the brain of the infant classic MSUD mice as well as the blood over the first few days of life. The rapid rise in brain leucine was accompanied by additional amino acid changes that suggested other large neutral amino acids were being prevented from entering the brain to establish normal concentrations. These amino acids included tyrosine and tryptophan, which are the precursors for the neurotransmitters dopamine and serotonin. This finding may help explain certain behavioral changes frequently seen in MSUD. Additionally, we found changes that suggested a dysfunction in brain energy metabolism.

The infant classic MSUD mice only survive for 2-3 days and are therefore very difficult to study. The intermediate MSUD mouse model, developed by the Pittsburg group was a stroke of genius, and has made possible the remainder of our studies. These remarkable mice have about 5% residual enzyme activity in the liver to metabolize branched chain amino acids (BCAA). The brain and all other organs have no detectable enzyme activity. About 90% of these mice survive weaning, but then become encephalopathic from BCAA accumulation, and all perish by 6-8 weeks of age. These mice provide an excellent model of MSUD in a fully developed animal that can then be studied while undergoing treatment trials. One of the most remarkable aspects of these animals is that they not only respond to a low BCAA diet, but provided with a choice of regular and no BCAA diet, they actually

regulate themselves. The proof is in the blood and brain. When the mice are allowed to choose their own diet, they actually consume mostly BCAA-free food, which results in reduced levels in brain and brings blood levels down to normal. When this was measured, it was shown that the mice consistently consume about 75% of the BCAA-free diet and 25% of the normal diet.

Through these studies with the MSUD mice, we gained the opportunity to try a novel treatment strategy. We showed that encephalopathic crisis could be caused in the mice on a low BCAA diet by introducing a high protein diet for 48 hours. We then introduced an "atypical" amino acid, Norleucine, into the diet. Norleucine competes with leucine for entry into the brain and prevents it from entering at the high rate normally seen in a crisis. We found that 5% norleucine by weight added to the high protein diet, prevented encephalopathy in all the mice for an additional 48 hours and for some more than 5 days. Analysis of the brains after this study showed that Norleucine had a more substantial impact on controlling the keto acid (alpha-ketoisocaproic acid) than lowered leucine levels. This finding strongly suggests that accumulation of the keto acid in MSUD is more toxic than leucine accumulation. Additional findings from transaminase deficient mice created by Dr. Sue Hutson support this conclusion. Transaminase (BCATm) deficient mice lack the ability to perform the first step in BCAA breakdown in all tissues outside the brain. These animals accumulate some of the highest levels of BCAA recorded for any organism, yet they do not develop brain injury. The ability to breakdown the BCAAs in the brain is still intact. Recent work with these mice shows a modest increase in brain BCAA, but no accumulation of the keto-acid and therefore no brain injury.

These findings have helped us to conceptualize a new area to focus on for potential treatment. The first step in BCAA breakdown is intact in MSUD but the second step, keto-acid breakdown, is blocked. Therefore, we find at least 2 opportunities to stop the mechanism of brain injury: 1) prevent BCAA accumulation, which is standard treatment through special low protein diet, and 2) prevent the BCAA from becoming a keto-acid (block the transaminase). Now it is time to go back to the mice and see if we can do this safely.

As I look back on the progress and the people gathered to study this complicated disease, I am encouraged by all the recent success and opportunities created. More recently, another MD/PhD student from Pennsylvania State University, Emilie Muelly, has embarked on the study of neurocognitive effects of MSUD through collaboration with the Clinic for Special Children. We look forward to Dr. Muelly's efforts to help understand MRI and behavioral changes associated with MSUD.

We are thankful to the MSUD family support group for continued encouragement, support and collaboration. Thank you! ■

## Low Protein Cooking Workshop and Family Dinner

Sponsored by Applied Nutrition

On August 3rd MSUD families from all over central Pennsylvania came together with Applied Nutrition to celebrate the launch of ***Homestyle Cooking: Recipes the Whole Family Will Enjoy***. The day was filled with cooking demonstrations and hands on activities featuring recipes from the new cookbook.



The workshop began with a step-by-step demonstration on how to make a delicious ***Vegetable Cheese Chowder*** by Glenda Groff. Glenda was an important part in creating the new low protein cookbook. She and other families from the Clinic for Special Children contributed most of the recipes, which were then tested and analyzed for leucine content and other nutrients by Applied Nutrition's dietitians. The event was an opportunity to showcase her recipes and provide additional tips to make low protein cooking and baking both fun and easy.



Next, Glenda led step-by-step demonstrations on how to make ***Oat Burgers***, low protein ***Hamburger Buns*** and ***Breadsticks***.

Everyone had an opportunity to make the bread recipes at one of six workshop stations. Here attendees had the learning opportunity to measure ingredients, learn how they interact with one another and watch the bread rise. Glenda also explained some

tricks of the trade, like adding extra wheat starch on a humid summer day to combat the moisture in the air.

Erica Novack, RD from Applied Nutrition, also led cooking demonstrations and a group activity. She began with a lesson on how to make ***Chocolate Zucchini Cake***, a wonderful recipe from the cookbook. As a dietitian, she pointed out nutrition tips and how wonderful adding vegetables to a dessert can be. The group loved this recipe and also enjoyed adding fruit into another new recipe from the book to make ***Fruit Cupcakes***. Each station measured, mixed and prepared their own batch of cupcakes and also made home-made frosting, which was used to top the cupcakes as part of the evening meal.

The evening portion of the event featured talks by Dr. Strauss and Dr. Morton from The Clinic for Special Children. Dr. Strauss discussed how the new Complex MSUD formulas were developed by Applied Nutrition in collaboration with The Clinic for Special Children to create a MSUD formula that would improve amino acid balance in the brain. He taught

the group about the importance of amino acids and how they work with one another. Dr. Morton addressed the group next about the importance of the low protein diet and eating smart to maintain a healthy weight. He urged everyone to take advantage of family gardens and fresh produce since fresh fruits and vegetables are generally low in protein, nutritious and filling. He pointed that instead of a tiny serving of potato chips that contains more fat and calories and will leave you hungry for more, you can enjoy nice helping of fresh cantaloupe that is quite filling and low in calories with the same amount of leucine.

Over 120 people attended and had a fun time sharing ideas, recipes and learning how to be more creative in low protein cooking. The event would not have been possible without the help of Glenda, the staff at the Clinic for Special Children and Applied Nutrition. All the dishes were a huge hit and were served as part of the main dinner. As a gift from Applied Nutrition, each participant received a can of ***Maddy's Homestyle Brownie Mix*** and a free copy of the new cookbook, ***Homestyle Cooking: Recipes the Whole Family Will Enjoy***.



My name is **Natalie Sullivan**.

## A PERSONAL STORY



I am 25 years old and I am a variant of Maple Syrup Urine Disease. I live in Toronto, Canada. I was diagnosed with the metabolic disorder at 5 months old. I have two amazing brothers and two wonderful parents. Growing up has been a struggle. Until the age of 3, I was in the hospital 36 times (averaging once a month). After that we stopped counting. I was a very sick baby. My mom, who is a physiotherapist, was asked to insert a N.G. tube when my levels became high and I wasn't vomiting. I hated having that tube being pushed through my nose and then later being pulled out! But this helped keep me at home rather than going to hospital. Sometimes I would rather have gone to the hospital!! My hospitalizations decreased to about two a year from the age of ten to twenty one. I haven't been admitted for over two years, which is a record. It remains a constant struggle of daily monitoring and diet management.

MSUD is a chronic disease that affects the whole family. My brothers Kevin and Keith were only two and five when I was born. I am sure that they missed us when I was in hospital. I remember bringing home little gifts for my brothers. It was a thank you for them because myself and either my mom or dad would be away from home, sometimes overnight or for several days. There were times when the car was packed and we were ready to go on holiday and I would be strapped into my car seat, my Mom would turn around, look at me, and announce "sorry guys but we can't go." I would be rushed down to the hospital usually with Dad and Mom to be home with my brothers. Kevin and Keith have always been very supportive of me and we continue to be very close.

It was very hard as a child growing up feeling different and feeling that I didn't belong. "Why am I so different?" was a question that I frequently asked. I felt different going out to restaurants with friends and always ordering the same things...salad and French fries. I felt different having to miss school in order to have my monthly blood work. I felt different not being able to participate in the hot lunch program at school. I wasn't able to commit to team sports because I didn't have enough energy. I did join the Girl Guides of Canada which I enjoyed for 9 years. One year I was on an overnight camping trip with the Guides. My cooler of food was mistakenly left out all night. When I got up in the morning we discovered that a raccoon helped himself to my low protein brownies and other food, but SURPRISE my formula was still there. I love to swim and was involved in swimming programs growing up. Today I am able to participate in our lake's annual mile swim. As I have grown older, I realize that I am not different but have a different set of challenges. Others have their own challenges.

My first MSUD symposium was in Montreal. I was about 4 or 5 years old. Since then I have enjoyed meeting other people with MSUD. We have maintained contact over the years and I consider them to be friends...friends with similar issues and understanding that no one else can really appreciate.

When I started high school I had set my goals, and am proud to say that I have achieved most of them. I graduated from high school, with an honorary award for college. I graduated from college and am now doing what I love the most. I work with pre-school children as well as after school-aged children. I have a nice fun group of friends. Right now I am happy doing what I do best. I have family and friends that understand and help me when I am sick as well as having a wonderful health care team.

I thank god that I have a wonderful family, amazing doctors and support group. So I was planning on having a liver transplant but because it's different in Canada I find myself waiting and waiting.

Love to all, Natalie Sullivan



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

## NORD Medical Foods Conference

The National Organization of Rare Diseases (NORD) hosted a Medical Foods Conference in Washington DC on February 10, 2011, to focus attention on an issue that is creating extreme hardship for patients and families affected by certain categories of rare diseases: access to medical foods. Representatives of the following stakeholder groups were invited to participate: patients, healthcare providers, professional medical societies, government agencies, insurers, and the medical foods industry. Sandy Bulcher and Amy Jones represented the MSUD Family Support Group at the conference.

NORD is committed to bringing this issue to resolution, and believes that many of you share that commitment. They are asking for representatives of the stakeholder groups, including support groups such as ours, to serve on a task force to drive this important work forward and periodically touch base with the larger group. Their summary follows:

### The Problem:

- There is no federal mandate regarding coverage of medical foods: 38 states have passed laws, but there is no consistency regarding diseases covered, ages of patients covered, whether mandate applies to private insurers and/or government. Also, some states cap benefits
- All 50 states now mandate expanded newborn screening but only 16 states mandate coverage for all screened IEMs; it is a moral imperative to ensure access to treatment for diseases identified through screening
- Lack of consistent definitions for medical foods
- Self-insured plans such as ERISA do not have to follow state mandates
- There is no clear diagnostic ICD codes for many of the diseases; (Several conference presenters mentioned that the coalition should address a range of diseases, not just PKU, including the organic acidemias, urea cycle disorders, and others)

### The Need:

- Average cost of medical foods varies, depending on disease, age of patient, etc., but is significantly higher than cost of regular food and frequently in the range of \$10,000 to \$12,000 per year; the cost of medical care is also increased if there is a delay in diagnosis / treatment for diseases requiring medical foods
- In the current economic environment, some state reimbursement services are being reduced
- Reimbursement is often linked to patient age, and compliance is significantly lower among older patients
- Access to treatment varies dramatically, depending upon state of residence or birth

### The Solution:

The solution will require action on several fronts, including some and possibly all of the following:

- ▶ Work together as coalition of stakeholders
- ▶ Address problems with HCPC codes
- ▶ Revisit current definition of medical foods used by FDA
- ▶ Support federal legislation
- ▶ Investigate possibility of getting medical foods defined as essential health benefit in healthcare reform

If you would like to volunteer for this cause which is so important to us all, please contact Sandy Bulcher at [dbulcher@aol.com](mailto:dbulcher@aol.com) or (614) 389-2739.

## Patient Safety and Formula

### Cambrooke Foods Inc.

A few months ago a pharmacy/supplier ordered PKU formula from Cambrooke Foods for a MSUD patient. The patient received the PKU formula and Mom gave her two pouches before they realized the problem. Fortunately the error was caught without any serious ill effects.

Cambrooke Foods uses Scancode shipping software which validates that what is shipped from the Cambrooke facility matches what is ordered. However, if the wrong product is ordered, that wrong product will ship from the facility.

The MSUD formula Camino pro drinks have a brown color scheme with a brown stripe that says MSUD Drink on the individual drink pouches. This same format appears on the cases. Based on this incident, Cambrooke has implemented additional security, a disorder-coded, two-inch wide strapping tape that is now adhered to the formula case boxes. (Tape is brown with repeated large white MSUD lettering.) If an error is made by a third party ordering system or a distributor, this alert tape should be seen by the end user.

Please contact Cambrooke Foods if you have any questions about your food or formula.

Call toll-free, (866) 4 LOW PRO / (866) 456-9776 or visit our web-site at

[www.cambrookefoods.com](http://www.cambrookefoods.com).



Sample of tape used.



## A Few Words From the Editor

Karen Dolins, EdD, RD, CDN  
Newsletter Editor

I received an email from a former student, now a registered dietitian and colleague, asking if we could talk about a personal issue. Her newborn daughter, now 10 days old, has been diagnosed with MSUD. For the first time in the 17 years since my daughter was born, someone I knew from my "other life" is joining our community. I was so thankful that I could tell her that her daughter would be OK, that by being diagnosed on the 8th day of life her prognosis was excellent, and that I'd be a key player in her support system.

As I review the content of this issue of our newsletter, I can see how far we've come. We now have the advantage of timely diagnosis, genetic screening, and collaboratives aimed at improving treatment. We have people who help us cope with changed dreams and navigating the future. We have the hope that current research will lead to even more improved treatment, and we have a strong community that keeps us all connected.

As you read through this issue I hope that you will feel buoyed by the advances made and the possibility of those to come. I hope you will be moved to advocate in whatever way you can, and that you will choose to share your family's story with us.  
Karen Dolins

## MSUD Symposium 2012

Save the Date:

# June 28-30

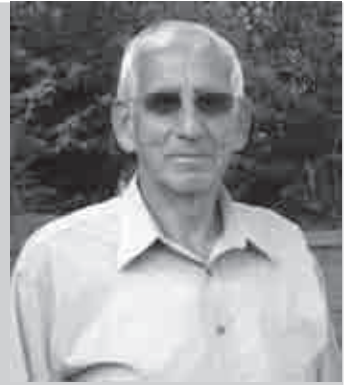
at the Embassy Suites- Airport  
Philadelphia, Pennsylvania

The hotel is conveniently located 1 mile from Philadelphia International Airport and offers free shuttle service to and from the airport. In addition, all hotel rooms are suites and include a living room and bedroom, plenty of space for the whole family. During your stay in Philadelphia, check out historic sights such as the Liberty Bell and Independence Hall.

Hope to see you there!  
Sandy Bulcher, Director  
MSUD Family Support Group

## Wayne Brubacher

President,  
MSUD Family Support Group  
Board of Directors



Wayne Brubacher is the President of our Board of Directors. He is the father of Monte (classic, 1965-1974) and Shayla (classic 41 years old), and husband of Joyce, former editor of this newsletter and active member of the support group. Wayne shares his thoughts on where our support group has been and where it is going.

Wayne is pleased to note the many advances made in MSUD treatment since the formation of our group in 1982. At that time, few doctors had even heard of MSUD. Parents needed a reliable source of information as well as support for the struggles of coping with this rare disease. The advent of the world wide web was instrumental in changing this. Now, physicians and families alike can search for the disease and quickly find information regarding the genetics of the disease, standardized treatment protocols, research, and our own website which gets up to 3500 hits a month. The website is accessed from people all over the world. We thank Eddie Wang for his tireless work in maintaining our website.

Due to the improved access to information, our group receives fewer requests for information. MSUD formulas are readily available, at least in the US. Advances such as branched-chain amino acid free TPN have improved care. Wayne feels that this has allowed our group to focus more on research. Our group supported the development of the MSUD mouse model, which allows researchers to study the effects of MSUD on the brain. With our support, Dr. Susan Hutson has conducted research using these mice to further our understanding. New treatment modalities are a major interest and the board reviews requests for support (see Dr. Zinnanti on cover).

Wayne notes that our group is only as powerful as its members, and hopes that all of you will do what you can to advocate and raise funds. ■



## FROM BABY TO COLLEGE STUDENT

### Joel Felsenstein's Story

By Linda Felsenstein

19 years ago, our state of blissful ignorance with regard to metabolic conditions was about to be shattered. Our seemingly healthy 3rd child - our only son Joel, was about to open our eyes to a way of life we could never have imagined.

To say the journey that ensued was without huge challenge would be stretching the truth somewhat, but in equal or even greater measure, the rewards, satisfaction and gratitude we have experienced and will continue to do so by having Joel in our lives is immeasurable. We have met many amazing people whose paths we would never have crossed had it not been for Joel. We have learned so much along the way and we hope we all, as a family, have grown in understanding others who face challenging situations. As the title of this article suggests (Joel chose the title), we have been blessed to be able to help Joel reach adulthood ready to embark on some level of independence which seemed an impossible dream when he was first diagnosed with MSUD.



19 years is an auspicious length of time for us. As Orthodox Jews, our lives very much revolve round the Jewish calendar. Birthdays and anniversaries are celebrated according to both the Jewish and regular calendar. Once every 19 years the dates of each calendar coincide, so a person's 19th/38th etc Jewish and regular birthday fall on the same day. This feeling of going "full circle" when Joel turned 19 prompted us to look back on his life and reflect upon how we arrived at where we are today. But let's go back to 19 years ago...

A perfect birth, a perfect baby and a smooth homecoming were but a fleeting hiatus before reality set in. Feeding deteriorated by day six and Joel could not be woken. Doctors, midwives, hospitals, tests, feeding tubes, IV's all appeared incredibly quickly and were destined to become integral parts of our lives. The doctor's words when trying to reassure us that Joel's deterioration was unlikely to be metabolic, I quote, "metabolic conditions are as rare as hen's teeth" had to be retracted five days later when indeed the results came back positive for MSUD. It was a partial relief when those five days of agony were over, knowing that at least we had a name for what was making Joel by now be on life support in the ICU. However, fear of the unknown at that point was in no short supply.



We will never be able to express our immense gratitude to Professor James Leonard (he was Doctor Leonard at the time), Marjorie Dixon, our amazing dietician and the rest of the team at Great Ormond Street Hospital in London. How do you ever thank people who literally save your child's life? Joel took seven long months to stabilize enough to be allowed home during which time my husband and I, together with supportive grandparents and friends juggled our two little daughters plus work commitments with being at Joel's bedside every single day. We learned how to tube feed, measure formula, balance protein with calories, pass an NG tube, take blood and deal with a range of therapists many of whom became an integral part of Joel's life.

Fast forward to school age by which time Joel had had several more hospital admissions, many health setbacks, various developmental crises and a swap from an NG tube to a gastrostomy. To be honest, school was never straightforward for Joel. He neither fell comfortably into special education nor into mainstream, but he never gave up and always tried his best. Each school we tried for Joel put themselves out so much to try to accommodate his needs even though at times this was an almost impossible task. Childhood illnesses, social difficulties, developmental delays and ever changing feeding regimes all contributed towards making it challenging for Joel to settle anywhere for long, although his final high school placement in a specialized unit within an Orthodox Jewish school helped him enormously both educationally and socially.

Joel was by now growing into a determined young man and this stood him in good stead through his teenage years. Life has definitely been at times an uphill battle for Joel but he has

*(Joel cont. on page 13)*



# MSUD Family Picnic



**John and Verna Martin** hosted a picnic at Leonard Harrison State Park in Wellsboro, PA on Saturday, June 11, 2011.

Attendees included individuals with MSUD spanning from 43 year old Lena Kurtz to babies Larry Nolt and Geneva Martin.

It was a beautiful, warm day, great for hiking and enjoying ice cream (regular and lo pro)!

*Babies Geneva Martin and Larry Nolt are pictured in their strollers at right.*



**Elan Geffen**, 27 years old, receives an adult leadership award from the Palm Beach County Business Leadership Network! Selection was based on his success at work, as well as community service activities. An awards luncheon was held at Northwood University in West Palm Beach, Florida.



## Hush Puppies

- 1/3 cup water
- 1 Tablespoon cornmeal
- 1 tablespoon finely chopped onion
- 1/2 cups Cambrooke Foods MixQuik

Mix ingredients together. Heat oil to 350 degrees and drop batter by teaspoonful into hot oil. Fry until they are golden brown, turning to brown evenly.

Per recipe	
Leucine (mg)	111
Protein (grams)	1.0
Calories	217

## Taco Soup

- 1 cup tomato sauce
  - 1 3/4 cups water
  - 1/4 cup chopped onions
  - 1 tablespoon butter
  - 1/4 cup uncooked low protein elbows
  - 2 tablespoons corn
  - 1 1/2 teaspoons taco seasoning
  - 3 corn chips
  - 1 tablespoon shredded low protein cheese
- Melt butter and sauté onion until tender. Add water and tomato sauce and bring to a boil. Add elbows, corn and taco seasoning and simmer 15 minutes. Ladle soup into bowl and top with crushed corn chips and cheese. Makes 2 serving servings.

	Leucine (mg)	Protein (grams)	Calories
Per recipe	232	2.7	332
Per serving	116	1.35	166

## Carrot Oven Fries

- 1 lb carrots cut into sticks or 1 pound baby carrots
- 1 teaspoon olive oil
- 1 packet (1oz) ranch dip mix
- 1 teaspoon cornstarch
- Cooking spray

Preheat oven to 400 degrees. In a bowl combine carrots together with olive oil, cornstarch and ranch seasoning and toss until well coated. Spray a cooking sheet generously with cooking spray. Arrange carrots in a single layer and bake for 25-30 minutes. Bake 35 minutes for extra crispy fries.

2 servings

	Leucine (mg)	Protein (grams)	Calories
Per recipe	216mg	3.6	260
Per serving	108mg	1.8	130

## Cabbage Soup

- 3 Tablespoons oil
- 1/2 cup celery, diced
- 1/2 cup onions, chopped
- Sauté until tender, add
- 4 cups water
- 2 packages George Washington's Brown Seasoning
- 1/2 cup diced carrots
- 3 cups cabbage, thinly sliced
- 1/2 teaspoon garlic powder
- 1 teaspoon Italian seasoning
- 2 cups chopped tomatoes
- 1/2 can Campbell's tomato soup, condensed
- 1/2 cup corn

Bring to a boil and simmer, covered, for 30 minutes. Serve.

	Leucine (mg)	Protein (grams)	Calories
Per recipe	646	6.8	610
Per serving	81	0.85	91

## Zucchini Rice Casserole

- 2 cups zucchini, sliced 1/2 inch thick
- 1/3 cup shredded low protein cheese
- 1/4 low protein rice
- 1/2 teaspoon salt
- 1/3 cup Campbell's Cream of mushroom soup, condensed
- 1/2 cup water
- 1/4 cup sliced mushrooms
- 1/8 teaspoon pepper

Arrange 1/3 of the zucchini slices in a buttered 1 quart casserole dish. Top with 2 tablespoons cheese and 2 tablespoons rice. Layer 2 tablespoons of cheese on top of rice and then another layer of zucchini. Layer the remaining 2 tablespoons of rice and arrange the remaining zucchini on top. In a small sauce pan combine soup, water, mushrooms, salt and pepper and bring to a boil. Spread over the top of the zucchini and sprinkle with the remaining tablespoon of cheese. Cover and bake at 350 degrees for 25 minutes. Uncover and bake 15-20 minutes or until rice is soft and zucchini is tender-crisp.

	Leucine (mg)	Protein (grams)	Calories
Per recipe	376	6.5	274

Please send recipes to  
Food News Editor

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## Cambrooke Foods – Summer 2011

Cambrooke Foods is bringing back a breakfast favorite - low protein bagels, including plain, onion and cinnamon raisin flavors. These bagels are single serving size and have a great chewy texture. Try them toasted with butter or your favorite jelly. Cambrooke is expanding their line of meat alternative products that contain DHA, to include our **Camburgers**. DHA is a polyunsaturated omega-3 fatty acid that is critical for healthy eye and brain development and has also been shown to support heart health. Numerous scientific studies confirm that everyone, from infants to adults, benefits from an adequate supply of DHA in the diet. Low protein diets are typically deficient of DHA due to the protein restrictions, so fortified "meat alternatives" are a tasty way to include DHA in your diet. Each serving of the **Camburgers, Tweekz, Corny Dogs, Brookelyn Dogs and Veggie Meatballs** has 32 mg of vegetarian sourced DHA.

Visit Cambrooke's Facebook and Twitter pages for information about Cambrooke Foods and metabolic disorders. Join interesting discussions and learn about managing your disorder.

Contact Cambrooke if you have not had the opportunity to try the Camino pro® formula line of ready-to-use drinks for MSUD. Available in Fruit Punch or Pina Colada flavors with 15 grams of protein per serving.

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

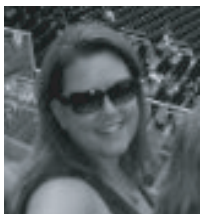
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## Journey of Purpose

Catherine A. Via Burzio, Education and Transition Consultant – **Viable Vocations**

The young adults walked into the conference session on a recent Thursday morning. Looking around the room each gravitated to familiar faces. The rowdy group of individuals age 17 and up could be likened to “Cheers - Where everybody knows your name and they’re always glad you came...” Loud hoots and shouts welcomed laughter and a few who prefer quiet tried to shush the crowd. Standing in the back of the room I couldn’t help but smile and soak in the networking, connections, and relationships evolving in front of me. I was overwhelmed. These are my kids. Not my own biological children, but I’ve known many of them since they were 7 or 8 years old, I know their families, I know their journeys...and they know mine. Once upon a time as a dancer/choreographer, I could count to 8 and 12 repeatedly while conducting bodies in motion. Blending movement with music to create a story or evoke an emotion was how I thought my days would be played out. The thought of karyotypes, molecular analysis, and data triangulation never entered my sphere of being. But during my second pregnancy, I began to learn about 4 letters – ACTG – and one number, 18, which would overpower but ultimately blend into my life and work.

My daughter Pauline was born on March 22, 1994. Two months prior to her birth, I had the first of four amniocentesis’ to determine if the baby I was carrying had a genetic condition causing the significant heart anomalies found two days earlier. The test showed she was a girl and she had Monosomy 18p-. We were flung into a subculture of medicine, terminology, statistics, and uncertainties. On that day, the obstetrician pulled us aside and said we needed to do two things - One, make sure our insurance was in order because this was our million dollar baby - and two, decide whether or not we would stay married. Having a child with special needs is extremely taxing financially, emotionally and spiritually. The divorce rate in the US is around 50%, but adding a child with special needs increases that rate to roughly 80%. Initially, Bret and I chose to walk, crawl and run through the journey together and it made the difference as we began to raise our family as a family. But one year after Pauline’s death, Bret chose to leave our family to pursue his life separate from ours.

Full of life, Pauline loved her friends, school, being in the junior high praise band at our church...she was a cool kid. But lest you think she was a perfect angel, she had her moments...like the multiple times she was caught pummeling her brothers. Or when she was found to be manipulating the school nurse into giving her chocolate from the private chocolate stash. Or the time she was caught writing something unkind about someone on the back of a bathroom stall! The punishment for that one was for the two of us to repaint the stall. Every time I go into Boyce Elementary school, I use that stall...makes me chuckle remembering.

Along with the diagnosis of 18p-, Pauline had multiple congenital heart anomalies (vsd, asd, pulmonary atresia, situs inversus totalis, etc...), a cleft lip and palate and GI issues. Upon her birth, we learned she had a low set ear nub on her left cheek and an underdeveloped left eye. This added to the cleft lip and palate is known as hemi-facial microsomia. Ashton was 4 when Pauline was born. He was an awesome big brother through Pauline’s entire odyssey. As she grew older, she would ask for him instead of us to meet a need. JB and August followed to round out our family.

Pauline required Early Intervention services including Physical, Occupational and Speech Therapies as well as education for our family in how to help her grow and interact effectively with her world. The people we met through the EI services became an integral part of our lives, continuing on even after the services ended. They showed us how to care for her and care for our family as a unit and the meaning of building a team to work with the singular goal of helping Pauline be the strongest member of our family and community she could be. This foundation gave her a huge leg up as she entered school based services at age 21/2 in an Early Childhood Special Education classroom. It was understood that Pauline would not move forward without a safety net of support and therefore, we were full members of the team...assisting with writing IEP’s, attending extra trainings and meetings...whatever it took to keep us moving forward together. We continued with the supportive environment through elementary school into middle school, but it took more effort to keep the focus on the family and not just status quo. We were blessed with individuals along the way who went out of their way to keep communications flowing and services happening to ensure Pauline would be educated well, with her peers.

Pauline entered Johns Hopkins Hospital on April 3, 2007 for a planned Fonteyn completion (heart surgery). She did well through the intense surgery, but on April 4, she suffered an unrelated severe brain bleed that ultimately caused her brain to swell and end her life at 4:15pm on Easter Sunday, April 8, 2007. Easter Sunday was fitting for a girl who knew her Creator and loved to worship Him. Pauline was able to be an organ donor and give life to two individuals. It has been over 4 years since Pauline’s death. Not a day or hour or sometimes a moment goes by that I don’t think of her. It is an excruciating road to crawl.

*(Journey cont. on page 13)*

(Joel from page 8)

never let this knock him for long. He has developed a healthy passion for everything electronic like most boys of his age, he has shown himself to be a talented musician playing both the drums and guitar (our house is definitely not a quiet one!)! and he has turned into a sociable guy as long as he feels he is in a secure environment with people who care for him. He is gaining independence all the time and is doing things we never thought he would achieve. We are so proud of him for taking the huge step of agreeing to go away to college soon and know that whatever obstacles come his way, he will be determined to overcome them.

From a baby whose prognosis was too dire for us to write about, to the young man we see today is worlds apart, and we thank G-d for providing us with the medical teams, appropriate dietary supplements and the strength to help him achieve the level of health and maturity that he has now reached. When we first met with the dietician, the diet seemed so baffling, especially as we would be balancing all Joel's metabolic needs alongside a kosher diet. However keeping a kosher diet also requires reading and analysing food labels, so we had a head start on this and now Joel is able to decipher ingredients and protein exchanges himself. In the last 3 months he has even begun to drink his formula and now hopes that the gastrostomy can be removed before he goes to college.

Joel has definitely kept us all busy since the day he was born and has given us plenty of frights over the years, but we love him for who he is and we admire him for his determined nature and insatiable appetite for life, both of which have brought him so far. We look forward to seeing him settle into college and are confident that the wonderful staff who will be caring for him, can help him reach his full potential. Meanwhile, we will look forward to him coming home each vacation as a mature and focused young man – something we could never have envisaged when he was a baby. ■

*Since writing the article, Joel has now successfully removed his gastrostomy and is taking all his supplements by mouth. This was especially pleasing to him as his ambition was to go to college without a tube. He has now completed two weeks at college so far and is doing really well. He and the staff are working very hard at learning how to keep him metabolically stable. Our dietician from London spent a day with the staff to train them in the art of cooking a low protein diet. They have been amazing at learning all about protein exchanges and calorie values of foods and they are introducing Joel to foods that he never previously tried. We hope that this move to college will help Joel gain vital skills to help him become more independent in the future.*

(Journey from page 12)

Pauline's death. Not a day or hour or sometimes a moment goes by that I don't think of her. It is an excruciating road to crawl. With 13 years of experience as an advocate for my daughter and training through Partners In Policymaking, in October of 2007 I was offered a position with the Parent Educational Advocacy Training Center (PEATC), Virginia's Parent Training and Information Center, as the Transition Coordinator. Through a Rehabilitation Services Administration grant, my role was to establish a Technical Assistance on Transition and the Rehabilitation Act (TATRA) center within PEATC. Over the next 3 1/2 years, I created a network of resources and information which breached the borders of Virginia and allowed me to connect with parents, educators and professionals around the country in order to support families and youth in transition with valid, current and relevant information. Through these networks I was introduced to Dr. Pam Leconte, Assistant Research Professor of Special Education and Disability Studies at The George Washington University. Within the Graduate School of Education and Human Development at GW, Dr. Leconte, is the program director for the Transition Special Education – Collaborative Vocational Evaluation Training (CVET) graduate program. Thanks to Dr. Leconte, I was able to secure a Fellowship in the CVET program allowing me to combine my "street smarts" as a parent advocate with academic "book smarts" in order to create the place I am now, completing my master's degree in order to better serve families and individuals in educational and employment transition.

In my current work, I help individuals with special needs identify vocational options by identifying and appraising their level of functioning in relation to vocational preparation and employment decision making. So now, instead of choreographing movement to music, I get to assist in the choreography of an individual's vocational journey, helping them create their own story based on their skills, interests, and values. The repetitious counting helps since data triangulation requires more than one source of information and flexibility is crucial since no two people, situations or assessments are alike. Established in March of this year, Viable Vocations has opened the opportunity for me to work in private practice supporting families and students through the maze of transition options and understanding the role of family in this process. As a trained professional, I understand that the technical components of the evaluation process only carry so far.

My life as my kids' mom allows me to work at a more core level of understanding, meeting families where they are and encouraging them forward. Working one on one with families, students, educators and professionals, my goal is to facilitate current choices to maximize future potential in the classroom, in the work place, in the community. What we are given to work with isn't wasted unless we let it go unused. Standing in the back of the conference room I felt overwhelmed looking at Pauline's peers, her "C18 buddies." I miss her, but what she taught me about living still has a purpose and I have the opportunity to share the next steps and transitions with that rowdy group of potential. ■

For information on how Catherine and Viable Vocations might serve you or your family, please contact us through [www.info.viablevocations.com](http://www.info.viablevocations.com) or [info@viablevocations.com](mailto:info@viablevocations.com) or 571-249-4902.

## Understanding Genetic Testing for MSUD



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The most important use for genetic testing for MSUD is to confirm a diagnosis in a child with a positive newborn screen result or positive biochemical testing. This is especially useful when biochemical results are indeterminate or in cases of intermittent MSUD, both of which can cause anxiety in parents. But genetic testing is helpful in other ways as well. Knowledge of the mutations in an affected individual allows for carrier testing in his or her family members. This knowledge also provides the option of prenatal testing or testing cord blood right after birth in a future pregnancy, allowing treatment to begin immediately if the baby is affected. Recent advances in genetic testing have allowed more and more people to identify the mutations in their family.

MSUD can be caused by mutations in any one of three different genes. These genes are named BCKDHA on chromosome number 19, BCKDHB on chromosome number 6, and DBT on chromosome number 1. An individual must have two mutations in one of these genes to have the disease, one from each parent. The mother and father, who each have only one mutation, are called carriers and they do not have any MSUD symptoms.

Genetic testing for MSUD starts with gene sequencing of the three genes. A blood or saliva sample is taken from the person to be tested and DNA is isolated. The DNA is then analyzed to determine the sequence, or spelling, of the three genes. Since we know what the sequences of the genes are when they are working correctly, the sequence of the DNA sample being analyzed can be compared to the correct sequence. Any disease-causing changes in the gene sequence that are identified are called mutations." If two mutations are identified, the individual has genetically confirmed MSUD.

Gene sequencing of the BCKDHA, BCKDHB, and DBT genes will identify both mutations in approximately 90% of individuals with biochemically diagnosed MSUD. But for the other 10% of individuals, only one or even no mutations are identified. Until recently, there was no further testing available to these individuals to help them identify both of their mutations. Now a test called a deletion/duplication array can be offered to the 10% of individuals who do not find two mutations by gene sequencing.

A deletion/duplication array looks at each of the genes to find large parts of the gene that are missing (deleted) or are present in too many copies (duplicated). While testing for deletions and duplications in the BCKDHA, BCKDHB, and DBT genes was possible in the past, it was difficult, time-consuming, and expensive, and did not always find every deletion or duplication. The deletion/duplication array currently available is easier to perform, quicker, and more accurate than the previous testing. So far in small studies, the deletion/duplication array has identified the "missing" mutations in almost everyone tested. Once both mutations are known in a family, then carrier testing, prenatal testing, and cord blood testing are all possible.

Still, in a few cases, genetic testing results may be indeterminate. These results are often called variants of unknown significance. For instance, a change may be found in the gene, but, because it has never been reported in anyone else, it is not known whether the change leads to MSUD or not. In these cases, it is helpful for the lab to have all the biochemical and clinical information about the person with MSUD so they can correlate that information to the genetic results. It may be helpful to test other family members to try to get more information about the change. The lab may look into large mutation databases to see if anyone anywhere in the world has seen the change before. When labs find unusual results, they will put those results into world-wide databases (without any identifying information about the patient or family) so that other people can learn from the results. When a variant of unknown significance is found, it can often delay the reporting of results while the lab tries to make sense of the change.

Genetic testing can be very complex and often raises many questions for individuals and their families. For more information on genetic testing for MSUD, ask your geneticist or genetic counselor. To find a genetic counselor near you, visit the National Society of Genetic Counselors website at [www.nsgc.org](http://www.nsgc.org). ■



Editor's Note: I met Andre at the Genetic Alliance meeting in June, where he was asked to present on the power of storytelling when living with disease. I hope you will all be encouraged to tell your own stories, amongst yourselves, to friends and family, and to other members of our support group via this newsletter.

## Telling Our Story: A Healing Experience

*I clearly remember the hot summer evening when I fell in love with story. The evening was warm with expectations of games of tag, orange Popsicles, and in the darkening night a thousand flashes of light from the fireflies. I noticed my parents and my neighbors gathering across the street in Hokies' kitchen. My childhood curiosity won the best of me, and I snuck into the house through the back door. I quickly went to my mother's side and sat silently. I anticipated her command to leave but no command came. I sat listening to their stories of World War II, the Depression, their childhoods, and their families.*

*I was too young at the time to convey what I learned about storytelling that night. However this experience and the many that followed that night were the foundation for a life-long love of storytelling.*

*When we tell our stories we convey not only the events of our life but also what those events mean to us. Imagine putting your hand in front of your eyes and slowly pulling it away. Initially, we only see our hand. However, as we pull our hand away we see not only our hand but also the rest of the room. The act of telling our story over a period of time is just like moving our hand away from our eyes. As we tell our story we broaden our vision and perspective and therefore the meaning of the event in our life. Also we grow in an awareness of new possibilities that suggest caring and healing actions we can do for ourselves and others.*

*Our stories are most healing when honestly told with humility. Most of us when we hear the word honesty think of telling the truth and not fibbing or lying. However, there is an additional understanding of the word honest. Honest in Latin breaks down into two words hon meaning honor and est meaning "self." In other words when we take the time to honestly tell our story we are honoring ourselves and our lives. Similarly the word humility derived from the Latin humilitas is often thought of as meaning meek, modest and timid. The Latin word humilitas with the root hum, meaning earth, has more of the sense of being grounded. So when we say we are telling our story with humility the connotation is that the story is grounded in our lives. So our stories are most healing when grounded in our feelings and thoughts, our struggles and victories and our ups-and-downs. When we honestly tell our story with humility there is an authenticity that honors both our life and those who hear our story.*

*To tell our story with honesty and humility takes time, patience and courage. We need to slow down from the busyness of our lives and patiently take the time to reflect and to allow our stories to unfold. Time for reflection may initially be difficult to integrate into our life but even a few minutes a day will make a difference. To do this we need courage. We initially need the courage to look at who we are, even though we may be afraid of what we may discover or that we might fall apart. We also need the courage to share who we are with others without hiding, detaching or hedging. What can help you in the processes is choosing a friend, relative or a support group that is trustworthy.*

*When we take the time to find and express our story we give ourselves to two gifts. The first is in the process of finding our story. We are giving ourselves the time to take care of ourselves, to discover who we are and what we need in the midst of a challenging time. Secondly, we are giving ourselves the opportunity to be heard and seen by others and in so doing allowing our selves to be supported. And finally in patiently taking the time to find and tell our story we support others in finding the courage and patience to face their life challenges.*

*As the night wore on the stories did not stop. I fell asleep first leaning against my mother's arm and then Hokie's arm. I felt safe and secure and knew I belonged. ■*

Andre Heuer D.Min. LICSW is a storyteller and a licensed clinical social worker. He has conducted trainings in the use of story for healing for war and torture victims in Liberia with the Center of Victims and in Thailand with SalusWorld. He also uses his storytelling approaches to work with individual's suffering acute and chronic illness. He has conducted workshops in storytelling and healing across the country. [andre@usfamily.net](mailto:andre@usfamily.net)



The following is a reprint of an article published in the Spring Issue of NIH's MedlinePlus:

## The Promise and Payoff of Rare Diseases Research, From NIH Director Dr. Francis S. Collins

Francis S. Collins, M.D., Ph.D., Director of the National Institutes of Health, led the successful effort to complete the Human Genome Project, a complex multidisciplinary scientific enterprise to map and sequence human DNA. He spoke recently with NIH MedlinePlus magazine about the increasing promise of genetics research to the investigation and diagnosis of rare diseases.

### Why should we focus on rare diseases when they affect so few people?

If you or your family were affected, it wouldn't be rare for you. And the study of rare diseases has taught us more than most people realize. Furthermore, the opportunities to capitalize on what we have learned so far have never been greater. If you care at all about biology and about understanding medicine, rare diseases are critical.

### How many rare diseases are there?

Altogether, rare diseases affect almost 25 million Americans. Worldwide, there are more than 6,000 that have an impact on people.

### How much progress has there been toward understanding rare diseases?

The good news is that we have learned a lot about the molecular basis of many of those that are caused by single genes that have gone awry. The bad news is that treatments are available for fewer than 200 of them at the present time.

(RareDiseases cont. on page 12)

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Also available



(RareDiseases from page 16)

**How much does the mapping of the human genome help?**

The Human Genome Project has provided many of the tools that have made it possible to reach our current understanding about the molecular causes of disease. But, I think it's fair to say that most of what we've learned from the genome project has not yet been applied. We want to accelerate that process. And that's one of my goals.

**What is the state of the art of genetics and disease now?**

The ability to identify the molecular basis of a disease, even a very rare one, has progressed rapidly. The challenge now is to develop clinical interventions in fewer than the 20 to 30 years it takes through traditional research methods.

**Have you an example of a disease on which there has been substantial progress?**

It has been just eight years since the cause of progeria, a rare childhood disease that causes rapid aging, was discovered in my lab. And we now have kids in clinical trials, some of them for more than two years. We were lucky here because the gene involved turned out to be one that we knew a lot about. And we were particularly lucky because that information suggested use of a drug that was developed for an entirely different reason; a "repurposing," if you will. That will happen from time to time, and we should not miss such opportunities.

*"The challenge is to cross the gulf between the molecular understanding we now have of thousands of diseases and treatment for them. This is where NIH plays a critical role in funding the necessary applied research."*

— NIH Director Dr. Francis Collins

**Any other diseases with similar progress?**

Yes. Research is showing significant potential for cystic fibrosis, sickle cell anemia, Niemann-Pick Disease Type C, and Fragile X syndrome.

**What does the research future hold?**

The challenge is to cross the gulf between the molecular understanding we now have of thousands of diseases and develop treatments for them. And this is where NIH can play a critical role in supporting the necessary translational research. ■

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## SECOND ANNUAL CHICAGO FUNDRAISER A SUCCESS!!!

By Karen Dolins

Friends and family gathered at the Dana Hotel and Spa in downtown Chicago on Saturday evening March 12th. The mood was festive, enhanced by the Roxx Box Band and food provided by Gene Kornota and Tony Klok of the Dana Hotel. Aided by a major donation by Dr. Lisa Ring, we cleared almost \$12,000 after expenses. Vitaflo and Cambrooke Foods also sponsored the event.

It was great to see MSUD families old and new along with friends and family who came to support our children. Hannah Dolins (17 classic) and Mackenzie O'Brien (13 classic) helped MC the raffle and silent auction.

Many thanks to Barbara Mudrick (mom of Dylan 3 years old, 2 years post-transplant) and my sister-in-law Karen Dolins (yes, we have the same name) who worked tirelessly to bring in donations and raffle items along with planning the event.

I was thrilled to have successfully bid on a quilt handmade by Wayne Brubacher's mother for her grandson, Joyce and Wayne's son Monte. Monte was a MSUD pioneer, the first in the world to be identified with MSUD through a state newborn screening test 12 days after his birth in 1965. Sadly, Monte died at the age of 9 from cerebral edema. Had he lived, he would have been 46 years old on March 12th, the day of our fundraiser. What was learned from his short stay on earth has helped improve the treatment of others with this disorder. Joyce and Wayne donated the quilt in the hope that the money raised will go towards research to help further improve treatment for the many facing the challenges of living with MSUD today.

Putting on the fundraiser was, I can't lie, a lot of work. The payback, though, was enormous and we got back much more than we put in. We are gratified to know that our work is helping to sponsor research into MSUD. We urge any of you who are considering putting together a fundraiser to take the plunge. You'll find it gratifying as well, as family and friends come together to help support our cause. ■

**Special Thanks**  
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for their generous donation  
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**Dr. William Zinnanti's**  
**research project.**

