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THE BATTLE FOR FORMULA LEGISLATION

Details

Written by Joyce Brubacher

Published: 20 July 2009

Sandy Bulcher

So much has happened this past year in terms of legislation for insurance coverage of formula that I hardly know where to start. I suppose I should start with a little history. Two years ago, metabolic families (primarily PKU) from across Ohio met to discuss introducing legislation that would require insurance companies to cover the cost of formulas and low protein foods.This was prompted by concern over decreased coverage of formula by the state and the ongoing expenses of paying for low protein foods. (Currently in Ohio all children and adults with PKU receive all of their formula free regardless of income. There is no state program for MSUD or other metabolic formulas.)

We metabolic families formed a group called the Ohio Coalition for PKU and Allied Disorders. Our first goal was to locate other families interested in our mission.We accomplished this by encouraging metabolic centers across Ohio to send out information about our group and its goals. Many families responded and expressed interest. Next we held several meetings to discuss how to approach the legislative process. A PKU family from the Dayton, Ohio area who had worked on Missouri legislation for formula coverage, proved to be a valuable resource. We developed a questionnaire and sent it out to all interested families. The goal of the questionnaire was to gather data about the metabolic families which could be used to support our case. We were in the process of gathering this data when, surprisingly, we learned that formula legislation had already been introduced.

My husband, Dave, was coming home from work one day last March of 1999 when he heard on the radio about the Bogan family fromDayton, Ohio and their plight to require their insurance company to cover the cost of their daughter Kaitlynn's formula. Kaitlynn Bogan was born severely allergic to food. She was diagnosed at six months with MSPI (milk, soy, protein intolerance).Kaitlynn had reacted severely to many different formulas and food. Her

reactions included severe abdominal pain, vomiting, diarrhea, rectal bleeding, skin rashes, and blisters. It was determined that the only formula Kaitlynn was able to tolerate was Neocate Infant Formula by SHS at a cost of $1,200 per month. The Bogan family, like so many of us, assumed that their insurance provider would cover the cost of Kaitlynn's formula - but they did not.

The Bogans eventually approached their local legislator, Representative Gene Krebs, who agreed to introduce a bill that would mandate insurance coverage of the formula.

I met with Representative Krebs in April of 1999 and encouraged him to broaden the language of House Bill 218 (the Bogan Baby Bill) to include metabolic formulas - low protein food is not included in this Bill. That is exactly what happened; he included the metabolic formulas. Little did I know then how far this journey would take me.

The Bill was initially introduced for consideration into the House Insurance Committee. Kaitlynn's mother, Lydia, provided powerful testimony about the severity of Kaitlynn's allergies, the expense of the formula, and the financial toll that it had taken on their family.

No insurance company testified against the Bill at these hearings; however, the Ohio Chamber Of Commerce did. The Chamber's goal is to protect the small businessman from rising insurance costs, which could potentially occur from the passage of a mandate. Incidentally, it was determined that our Bill would cause an increase of approximately one cent per year on the average family's insurance policy.

Thankfully HB 218 passed the House Insurance Committee by a large margin, 16 to 4. We were thrilled and believed naively that this process wouldn't be so tough after all.The next step would be a vote from the full House.

We soon learned that the Bill needed the approval of the Rules and Reference Committee. This Committee decides which bills move to the full House for a vote.The residing chairperson of this committee and current Speaker of The House is Jo Ann Davidson.

Lydia Bogan and I called many families from across Ohio whose children drink special formulas, and encouraged them to call Speaker Davidson's office and express their support of House Bill 218. Within two days, her office received over 500 phone calls. We learned later that this many inquiries about one bill is very unusual. Still, the Bill did not move to the full House.

Through the summer of 1999, while the legislators were in recess, Representative Krebs held several meetings with key parents to develop strategies to move the Bill on to the full House for a vote. Also during the summer months, I created a data base of parents and interested professionals so that I could easily communicate via e-mail. Many families contacted their local legislators and informed them about HB 218.We also wrote letters to the Ohio Chamber of Commerce and several other business organizations who continued to express opposition to the Bill.

Fall arrived and we held a rally at the Ohio State House in Columbus. Families with children with severe food allergies, malabsorption syndrome, and metabolic disorders attended, as well as several PKU adults. After the rally, families met with legislators to educate them

about the Bill. The rally received coverage from radio, TV, and newspapers. The public response was overwhelmingly in support of this Bill.Still the Rules and Reference Committee refused to move HB 218 to the full House. It seemed there was always "bigger and more important" issues the legislators were attending to.

In November, I met with Speaker Davidson regarding House Bill 218. It was a valuable meeting. She learned the "human" side of the story. I was able to educate her about MSUD and how vitally important the formula is to keep Jordan healthy.

During the fall and winter months, Lydia Bogan began her quest for more media attention on House Bill 218. She contacted many national programs. The interest was overwhelming. We also decided that it was time to name our group.After some deliberation, we decided on MAMA (Mothers Aligned for Medical Advocacy). It has proven to be a good choice, as it is easy to remember and recognize.

During the fall, we were contacted by a group of high school students interested in our Bill. Led by Representative Kreb's daughter, Alania Krebs, this group developed a web site to educate persons about the Bill and they continue to hold fundraisers to help families who can't afford life saving formulas. We were touched by their generosity and sensitivity to our issue.

We were hopeful that the House would vote on our Bill after the holidays. Still the Rules and Reference Committee did not move the Bill. Lydia Bogan decided to take our issue to a higher level. In January of this year, she contacted the campaign managers of the presidential candidates and began educating them about HB 218. Quite honestly, I never expected them to be very interested, but I was wrong.

During the month of February, when the presidential candidates were campaigning in Ohio for the Primaries, we were able to talk with all of them regarding our Bill. Lydia talked on the phone to Bill Bradley about HB 218 and part of her conversation aired on local TV. Several mothers spoke to Vice President Al Gore at a rally in Dayton, Ohio. Four families attended a McCain Town Hall Meeting where Lydia had the opportunity to discuss HB 218. Lydia met personally with Pat Buchanan and shared her concern about the lack of insurance coverage of special formulas.

In late February, five families from the MAMA organization met privately with Governor George W. Bush. He, like the other candidates, expressed his support for our Bill. That day Governor Bush spoke with Ohio Governor Bob Taft and the Speaker of the House JoAnn Davidson, and encouraged them to move this Bill forward. Most recently, several mothers received personal invitations to attend a Gore Town Hall Meeting. Following the meeting, we approached Vice President Al Gore about the Bill. As soon as we mentioned "formula," he replied, "Oh yes, the Bogan Baby Bill."

Clearly this issue has received more interest and attention than I ever dreamed possible, largely because of the persistence of Lydia Bogan. The need for federal legislation to mandate insurance coverage of special formulas is real, and the next President of the U.S. will be well aware of that. Incidentally, the MAMA organization is a non-political group. We are seeking support of this issue from both the Democratic and Republican Party.

Lydia's quest for media attention for our Bill continued, and in March, CNN ran a story about the Bogan family. As I write this, the Rules and Reference Committee still has not moved HB 218. I feel confident, however, that the MAMA group has made a difference, and the legislators are well aware of how important this issue is to us.

I believe that we will see the Bogan Baby Bill become law before the year is over. Then we plan to use that momentum to take us straight to Washington D.C. for federal legislation that would require insurance coverage of special formulas.

Update May 21:
Unfortunately, because of pressure from the opposition, Speaker Davidson has decided not to move the Ohio formula Bill through the House during this session. This means we have to start all over again.

While I am discouraged about this, I remain optimistic that the MAMA organization has made a HUGE impact, and the second time around will be easier. We have term limits in Ohio which means many legislators have completed their terms, and new persons will be elected in November. We will have a new Speaker of the House, who is well-educated about this Bill and more supportative. In addition, we will need to find a new sponsor for HB 218 because Representative Krebs will complete his term at the end of this year.

Several legislators have expressed interest already.It has been a long tough process, but I remain confident that the legislators will realize that passing this Bill into law is the right thing to do. The MAMA organization will continue to push for a federal mandate that would require insurance companies to cover the cost of special formulas.

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THE LOWDOWN ON LEUCINE - GRAMS, MILLIGRAMS, AND EXCHANGES

Details

Written by Joyce Brubacher

Published: 20 July 2009

Rani H. Singh, Ph.D., R.D., L.D. and Pennie A.Graham, R.N.
Dr. Randi Singh is a nutritionist at Emory University in Atlanta, Georgia, and a nutritional advisor for our support group.Dr. Singh submitted the following article on a subject we families deal with every day. She offers advice and the promise of more help on the way.

It can be a major challenge to successfully meet the special dietary needs of persons with MSUD while maintaining both food variety and satisfaction. It requires an understanding of the amount of leucine in different foods, how to calculate the amount of leucine eaten, and how to use this information to make food choices while remaining within recommended daily intake ranges.Another challenge is learning how to include foods with unknown leucine values. At times, it seems to require an advanced degree in mathematics to determine serving sizes and food options. This tedious approach to daily food intake can discourage one from comparing foods, adding new foods, and correctly calculating leucine intake.

In a recent random survey of the MSUD population in a number of states, we found that several different systems were being used to calculate leucine. Many families counted total protein grams (g). Others counted milligrams (mg) of leucine. Still others used an exchange system in which 20 to 30 mg of leucine was counted as one leucine exchange. While all of these systems can work well when managed correctly, we found that little agreement exists about which system works the best and is least cumbersome in the hectic pace of daily living.

When children are young and parents control the diet, counting leucine in milligrams seems the preferred and easiest way to keep track of dietary leucine intake. As children grow and are exposed to food from outside sources, it becomes more difficult to determine exactly how much leucine is present in the meals they eat at daycare, school and away from home.

As children mature and begin to take more control of their own dietary choices, they may want to add new foods to their diet. More complex math is required to figure protein grams and leucine milligrams from the available information about foods on the market. This is frequently a stumbling block to making good dietary decisions for themselves.

We have a special metabolic camp held each summer for persons with PKU and MSUD 10 years and older. At the camp, have been working with several management systems to devise a simple, easy way to insure dietary compliance while providing the campers with a better understanding of their dietary options and giving them a sense of mastery over the food related decisions in their lives.

The program we utilize is based on two basic principles:
1) 30 mg of leucine = 1 leucine exchange.
2) Leucine = approx. 6% protein (an average determined by the 3.5% to 8.5% leucine/protein ratio in different foods).
Understanding these two principles allows children to quickly estimate leucine values and make healthy dietary decisions.
For example, a 1 oz. bag of potato chips has 2 grams of protein. To determine the leucine value, the child can simply follow these steps:

1. 1. Convert grams to milligrams by moving the decimal point 3 places to the right:
   2.0 g = 2000.0 mg protein
2. 2. Multiply the mg by 6%:
   2000 mg x .06 = 120 mg
3. 3. Divide the mg of leucine by 30 to find the number of exchanges:
   120 mg ?30 = 4 exchanges

Note: An even easier way to estimate leucine is to remember that 1 g of protein = approximately 2 leucine exchanges.

Having determined the exchange value, the child can decide whether or not to spend 4 exchanges of their daily leucine intake on a bag of chips.

Response to this system, while still preliminary, has been encouraging. The campers have indicated they feel comfortable with this system and are more readily able to make the correct food choices for themselves. They also appear to better understand how their choices impact their overall health.

As a result of this positive response, we are now developing a pocket-sized booklet which lists the amounts of protein (in grams), leucine (in milligrams) and the number of leucine exchanges for many foods. These amounts are converted into household measures for easy use. The source of this information is a detailed computer data base (Amino Acid Analyzer, Ross Nutritionals). By including all three methods of measurement, we hope that this booklet will be a tool to assist all families with MSUD, regardless which system they currently use to calculate leucine intake.

In the near future, we will be surveying dietitians and consumers about the food list to confirm the results we have already seen in our camp population. Once this is completed, and all comments and suggestions have been included, we hope to distribute this booklet to the general MSUD population - possibly by the end of this year. It is our hope that this new, simpler approach to the dietary challenges of living with MSUD will help families implement more variety, satisfaction and control into their food choices.

If you would like to provide input at this stage, please e-mail me, Dr. Singh, at: rhs@rw.ped.emory.edu. We welcome your comments and suggestions.

NOTES - "CAN GENE THERAPY CURE THIS CHILD?"

Details

Written by Joyce Brubacher

Published: 20 July 2009

Persons interested in the gene repair therapy that Dr. Blaese spoke to us about at Symposium '98 will want to read the article in the May issue of *Fortune* magazine titled, "Can Gene Therapy Cure This child?" The article reviews the Kimeragen company's struggle to break into the gene therapy business. It chronicles the problems involved in bringing the therapy to human trials.

The Floyd Martin children from Pennsylvania with Crigler-Najjar Syndrome, are scheduled to be the first to receive the new gene repair therapy.They are featured in the article with a full page photograph of Amy Martin on the first page of the article.

The article is lengthy and very interesting, although MSUD is not mentioned. At Symposium '98 Dr. Blaese told us that children with Mennonite classic MSUD would probably be next in line for trials after the Crigler experiments. This was expected to happen within a couple of years. This article reveals why this hasn't happened yet.The first human trials using this gene repair therapy will be conducted in connection with Dr. Morton's Clinic For Special Children in Strasburg, Pennsylvania. Dr. Blaese will not be able to attend Symposium 2000, so Dr. Holmes Morton will give us an update on the gene repair therapy at that time.

NOTES - ADVOCATING NEWBORN SCREENING

Details

Written by Joyce Brubacher

Published: 20 July 2009

If you don't have e-mail, you may not know about the *Tyler for Life Foundation*. This new foundation is a newborn screening awareness organization in Georgia. I am on the board as directors of the Tandem Mass Spectrometry Disorders Division. My job, with the help of others, is to see that the 3.7 million babies born each year in the U.S. get screened with the very effective tandem mass spectrometry technology. Tragically, 2,700 children die each year in the U.S. or are brain damaged because someone did not know they should have a $20 newborn screening test [which includes MSUD]. We plan to work on the international level in a few years.

We can't do it alone. Please contact us to see what you can do to help educate the world and educate your community. The world has GOT to know that newborn screening for many of these disorders EXISTS! You can get newsletters and information by contacting *Tyler for Life* online or off line.

- Wendy Nawn
Wendy Nawn, MCAD (610) 251-9876
Tera Mize, galactosemia, President (770) 947-3638
Tyler for Life Foundation, Inc.
6340 Holborne Lane
Douglasville, GA 30134-4023
www.tylerforlife.com

NOTES - CONGRATULATIONS TO THE NEWLY MARRIED

Details

Written by Joyce Brubacher

Published: 20 July 2009

The families who attended Symposium '98 in Lancaster, Pennsylvania, will remember the Personal Perspective Panel. The young adults on the panel answered questions from the audience. Jaime Ness from Wisconsin, Michael Woorman from Maryland and Sharlene Balinsky from Montreal, Quebec were all on that panel. Now their lives have taken on a new dimension.

JAIME & BRIAN

On Saturday, February 26, 2000, Jaime married Brian Hamilton at the Lodi United Methodist Church in Lodi, Wisconsin. Jaime is 22 and has classic MSUD. She is the daughter of Christine and James Ness from Lodi, Wisconsin. Brian is the son of Karen & William Hamilton from Columbus, Wisconsin.

Brian and Jaime had five attending couples (including her sister Vikki, and Brian's twin brother Brice and his sister Bobbie). Jaime's mother, Christine, reports: "The weather was very unusual and warm for February with a thunderstorm about one hour before the wedding, and temperatures in the low 60's. The weather was one of the biggest stresses for planning, because we could have had rain, sleet, snow or a blizzard with very cold temps. So God was smiling on Jaime's special day and allowed her to have beautiful weather with no worries of bad roads."

One unforgettable event of the day was when their limousine driver was arrested by the police on the way from the church to their reception in Waunakee. The photographer following them took over driving the limousine while his wife got all the action on video. But the police had arrested the wrong driver! So the limousine company offered Brian and Jaime another opportunity to use their service for any special event at no cost.

Christine says the couple may consider a limousine ride in June to bring a newborn baby home from the hospital. Jaime is expecting to deliver a baby around the middle of June. She is disappointed that she will not be able to attend Symposium 2000. But the disappointment is overshadowed by her anticipation of becoming a mother. The pregnancy has not been easy, but all seems to be well as of the middle of May. Jaime is eager to share her experience with the other families. There will probably be news to announce at the Symposium. Pray that all will go well for Jaime and the baby.

NOTE: Jaime gave birth to a healthy baby girl on June 19th. Mother and daughter are fine.

SHARLENE & MICHAEL

Sharlene Balinsky and Michael Woorman, both 25 years old, first met in 1988 at the Symposium in Lancaster, Pennsylvania. They corresponded and kept in touch over the years until they once again met in Pennsylvania, at the 1998 symposium and have been together ever since. On Valentines day l999, Michael proposed, and they became engaged. On April 2, 2000, they tied the knot in a traditional ceremony at the Shaar Hashomayim synagogue in Montreal.

Sharlene has classic MSUD and Michael has a variant type of MSUD. We believe they are the first couple with MSUD to marry.

The couple resides in Maryland where Michael works as a consultant and salesman in a carpet shop. Sharlene continues to work with preschoolers.

NOTES - PARENT CONFERENCE IN MICHIGAN

Details

Written by Joyce Brubacher

Published: 20 July 2009

Shayla and I just came back from attending the 16th parent conference put on by the Association for Neuro-Metabolic Disorders in Ann Arbor, Michigan on May 20. The conference was entitled *Parent Forum: Experiences With Metabolic Disorders.*

Eight families with children with MSUD attended. The families shared in an informal discussion group during the morning. This was a lively time, with good-natured banter along with serious discussions on diets and sharing the progress of each of our children. The families did not have their younger children along, but Shayla enjoyed being with Amy Whitfield, 23, and Alana Moceri, 16.

Shayla appreciated the talk and slides on "Blood Collection Issues in Metabolic Disorders" given by Denise Pleger, RN. Shayla takes her own blood and learned some helpful hints.She enjoyed viewing the slides showing what happens to the blood dots sent to the laboratory.

As a teen presenter, Alana Moceri shared how she felt about having MSUD. She encouraged parents to give lots of praise and reinforcement to their children which motivates them to reach their goals. Her dream is to build her own day-care center.

- *Joyce Brubacher* (Copied from the Editorial)

NOTES - NEWBORN SCREENING NOTE FROM JOYCE

Details

Written by Joyce Brubacher

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Luke and Laura, a young married couple from our area of Indiana, knew they could have a child with MSUD. They both had relatives with children with MSUD. They were tested and found to be carriers of the Mennonite mutation for MSUD.

When Laura became pregnant, we began planning to test their newborn immediately after birth. Arrangements were made to send blood to the Clinic For Special Children in Pennsylvania. We also sent a cheek swab to a laboratory in Columbus, Missouri for DNA testing. The usual state screening tests were sent to the state laboratory in Indianapolis.

The test results from blood drawn at 14 hours were available from the Clinic the next day when the baby was 48 hours old. The results of the DNA tests were called to them a few hours later. The infant was not even a carrier - totally free of MSUD. The state tests, showing a slightly (false) positive reaction, were reported almost 2 weeks later.

Our youngest son Troy and wife Carolyn had his first child on May 19th, our first granddaughter.It will be one week tomorrow since her newborn screening test was taken, and we still do not have the results from the state. Although we did not have to rely on the state tests, I am appalled at the slow response. This is not acceptable.

A parent group is advocating TMS screening for Indiana. Hopefully the whole screening program will be improved soon. What is the situation in your state?

- Joyce Brubacher

NEW FOODS

Details

Written by Joyce Brubacher

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Scientific Hospital Supplies (SHS) is marketing a low protein cereal, Breakfast Cereal Loops. They taste like Honey Nut Cheerios and not at all like a low protein food. The cost is $6 per box or $5 per box if you buy 4 or more boxes. There are 13-one cup servings in a box and 1 cup has 3.4 mg leucine, 0.14 g protein and 110 calories.

Shipping and handling charges for less than a $25 order is $5. There is no S&H charge for orders over $25. To order, call: 888-567-7646 or order from:

SHS North America, P.O. Box 117, Gaithersburg, MD 20884-0117

Dietary Specialties is now marketing several fully prepared low protein foods. Each product, except the Macaroni and Cheese, arrives frozen, shipped in dry ice.

- *Imitation Macaroni and Cheese*- Makes 3 servings. $4.99 per pkg. or 12 pkg. in a case for $55.08. One cup serving has 92 mg leucine.
- *Wheat Starch Bread*- 14 oz. loaf for $5.59 or 8 loaves in a case for $39.92. Approximately 16 slices per loaf and 20 mg leucine for 2 slices.
- "*Rye-Style" Wheat Starch Bread with caraway seeds*- 14 oz. loaf for same price as bread above. Approximately 16 slices and 27 mg leucine for 2 slices.
- *Dinner Rolls*- 12 oz. pkg. for $4.59 or case of 11 pkg. for $44.99. 6 rolls per pkg. and 23 mg leucine per roll.
- *Pizza with Imitation Cheese Topping*- 8" pizza for $6.39 or case of 9 pizzas for $53.91. 4 servings per pizza and 107 mg leucine per serving.
- *Pizza shells*- 8" shell with 2 per pkg. for $4.39 or case of 6 pkg. $23.94. A serving (? shell) is 25 mg leucine.
- "*America -Style" Singles Imitation Cheese*- 10.7 oz. pkg. is $2.99 or case of 12 pkg. is $32.28. 16 slices per pkg. and 52 mg leucine per slice.

The newest item is a DariFree Beverage Mix. For more information or to make an order, call: 888-636-8123 or send to: Dietary Specialties, 865 Centennial Ave., Piscataway, NJ 08854

Cambrooke Foods is the new company introduced in the last issue of the Newsletter. There have been unforseen delays in getting these new foods marketed. They include bagels, energy bars, pizza crusts, Caramel Tapioca Cakes (comparable to rice cakes) and Creamy Hot Cinnamon Cereal (comparable to flavored cream of wheat). Keep checking the web site to learn when these new, tasty products become available: www.cambrookefoods.com.

We try our best to see that the information in this *Food News* section is accurate. The amino acid values listed with the recipes are provided by nutritionists from Emory University. We are not responsible for typographical or other errors. Contact your clinic before making diet changes.

FAMILY HISTORIES - KYRA VAN SCHAIK, A LITTLE DUTCH GIRL

Details

Written by Joyce Brubacher

Published: 20 July 2009

Marian van Schaik

First I'd like to introduce myself. I am Marian van Schaik. I'm married to Marcel, and our daughter, Kyra, has MSUD. At this moment we are expecting our second child and we already know that this baby does not have MSUD, which is a big relief. We are Dutch and lived in the Netherlands until May 1999. We moved to Auburn Hills, Michigan, last summer. I would like to share our story about Kyra with all the people who read the MSUD Newsletter.

On Sunday, October 26, 1996, after an absolutely perfect pregnancy, Kyra was born. She was a beautiful baby, and we were in seventh heaven. The only thing that wasn't going so well was feeding. Kyra just refused to drink and would keep her mouth tightly closed when we tried to feed her. But when we'd offer her a finger, it did show that her sucking reflex was functioning normally.

After three days, we noticed that Kyra would stretch her body - her spine in a hollow position and her head backwards - when changing diapers and dressing her. She was also moaning and crying with a strange, high-pitched voice. She would still drink only a few minutes at a time, and on Thursday (the 4th day) she wouldn't drink at all. Together with our midwife, we decided to call our family doctor on Friday morning.After examining her, he said he didn't know what was wrong with her and decided to call the pediatrician at the local hospital. The pediatrician said we could come in right away, and that he'd probably hospitalize Kyra.

After the pediatrician had examined Kyra, he said, "I don't know what is wrong with this baby, but it's not good at all." It turned our world upside down. In the meantime, due to being undressed for a while, (although under a red lamp), Kyra's body temperature had dropped to 95 degrees. So the doctor decided to put her in an incubator. It had to be kept quite warm (107 degrees) to keep Kyra's temperature from dropping again. She also had an IV because she was dehydrated, and she was being fed by a tube in her nose.

Because the pediatrician didn't know what was wrong with her, he collected some blood and urine samples from Kyra, and sent them to the university hospital in Amsterdam.By Tuesday evening, they could tell us the name of the disease our daughter was suffering from - MSUD. The only thing the pediatrician could tell us at that moment was that this is a metabolic disease, and that it affected the breakdown of protein in the body. Kyra was now nine days old.

The next day, Kyra, who was still in an incubator, was taken to the university hospital in Amsterdam by ambulance. I was allowed to ride with her, and, although I knew exactly how to drive to this hospital, I didn't recognize the route at all. At the hospital everything was prepared to put Kyra on a restricted protein diet. MSUD Diet Powder was ordered, and we were informed by a nutritional advisor as to what and how to feed her. From that moment, her meals were the MSUD formula mixed with water and just a little bit of baby milk powder equivalent to 2 mg of protein per day.

Ten days later Kyra's leucine levels were still very high, so the doctor decided to give her hemodialysis. She had to stay on the machine for two days, which finally brought her leucine value down to an acceptable level. In a few days Kyra's appetite was improving and she even started to cry if we didn't feed her in time. The feeding tube was removed, and we started feeding her by bottle. Finally we dared to hope everything would be ok with our little girl.

On December 2, 1996, Kyra came home from the hospital. She was fine for about two weeks. Then another problem arose; Kyra was anemic. When we woke up in the morning, I would touch her first to see if she was still alive, because she looked very pale. We went to see the doctor, and he decided Kyra needed a blood transfusion. This happened every other week. Between times, Kyra would do well for a few days then start vomiting after or during feeding.

We'd wait for about a day or two to see if things would get better, and just when we decided this couldn't go on, Kyra would be back to "normal" for a few days.She also was developing a diaper rash that didn't go away. Whatever I tried, it seemed to get worse.

On January 14, 1997, Kyra was hospitalized again. She refused to drink her formula, and her rash was really bad now. It looked as if she had been sitting in boiling water, and she was in a lot of pain. Even her face, arms and legs were turning red, and the inside of her mouth was swollen.The pediatrician at first thought it was a virus, but the dermatologist could tell us right away that this was being caused by isoleucine levels that were too low. That meant we had to add isoleucine to her formula and go back to tube feeding her again. Although still on a feeding tube after two weeks of hospitalization, we could take Kyra home again. We all thought that, being at home, Kyra's condition would improve quickly. That was a big mistake.

Sometimes it took Kyra 1, hours to finish half of her bottle, which contained only 85 cc! Of course she still had the feeding tube, but we tried to use it as little as possible, because we thought it would be best if she'd drink as much as possible by herself. Things got worse and worse, and, finally, on February 6, 1997, we decided to have Kyra hospitalized again to find out why she refused to drink. She also was still anemic and needed transfusions.

This time Kyra was in the hospital for six weeks, and nobody could tell us why she was vomiting and still needed to be tube fed. All kinds of medications were being given to her, but nothing really helped. They even told us at one period of time that it was a psychological thing; the fact that Kyra never had a pleasant feeling when eating caused her to refuse food.

I won't bother you with all the little and big things that went wrong during that period of time, but I can tell you that our faith in, and respect for, the doctors in that hospital faded more and more every day. Finally my husband, Marcel, decided to contact the MSUD Family Support Group while on a business trip in the U.S. This was in the first week of March 1997. He talked to Joyce Brubacher, and he later told me how wonderful it was to talk to someone without any medical background, who completely understood what we were going through.

Joyce gave him the number of the Clinic For Special Children, and, much to his surprise, he could talk to Dr. Holmes Morton right away. From the little information Marcel could give Dr. Morton, he could tell Marcel that Kyra was suffering from malnutrition. This meant we needed to increase her leucine intake drastically.

Marcel called me right away to tell me about what he'd learned, and wanted me to tell the pediatricians in Amsterdam about this (we thought) wonderful news. To my big surprise, the pediatrician I talked to reacted very skeptical, but he was willing to give it a try. In less than a week her leucine/protein intake was increased more than 10 times, and we could clearly see an improvement. One week later we left the hospital. We felt that what was being done there for Kyra, we could do at home too.

After talking to Dr. Morton several times, we had so much confidence in him, we wanted to meet him personally. This soon became possible. Marcel had to go on another business trip to the U.S. by the end of March 1997. At first, Marcel planned to go alone, but Dr. Morton suggested he take Kyra and myself along. Fortunately, we were able (with assistance from our local doctor in Amsterdam) to convince our insurance company that this trip would be very important. So Kyra and I joined him.After his work was done in Detroit, (where we were kindly invited to stay with friends), we flew to Philadelphia and then visited the Clinic.

Dr. Morton had planned to hospitalize Kyra for three days in Lancaster for an MRI and to regulate her leucine intake. What struck me most was that, when we arrived on Wednesday, Dr. Morton said, "Tomorrow I'm going to take out that feeding tube!" And that's exactly what he did. By Thursday afternoon Kyra was drinking her formula by herself!

By that time, she was 5 months old and had been hospitalized a total of 13 weeks. Her MRI, taken immediately after we arrived on Wednesday, showed the brain development of a six week old baby, caused by not getting enough protein.

Although it took some time, everything went well. We decided, after talking with Dr. Morton and our doctor in the Netherlands, that Dr. Morton would be our consultant. The university hospital in Amsterdam would determine the amino acid levels, and fax the results to the Clinic. Then we would call Dr. Morton for advice on how to change Kyra's diet.

Kyra started to develop at a very rapid rate, and by the time she had her first birthday, she had almost reached normal development for her age. This was confirmed by Dr. Morton in his Clinic, on a visit in October linked to our vacation on the west coast. Kyra hasn't been

hospitalized since, and is doing wonderful now. She is three years old and looks and acts just like any other ordinary child her age.

The decision to move to the U.S. last year, was greatly influenced by the fact that Kyra has MSUD, and that there is more knowledge about treatment here. To put that into perspective, the doctor who treated Kyra in the Netherlands had his last case almost 30 years ago. When Kyra was born, she was one out of three children with MSUD in the Netherlands.

I am convinced that if it wasn't for the MSUD Support Group at first, and, of course, Dr. Morton, who helped us through these years and still does, Kyra wouldn't have made it to her first birthday.

FAMILY HISTORIES - NICK AT 21

Details

Written by Joyce Brubacher

Published: 20 July 2009

In 1982, Nick Lovrin and his mother and father flew from California to Indiana to attend the very first MSUD Symposium. Nick was only 3 years old, and I remember him as a cute pleasant, healthy-looking, little boy. He is now a young man, still living in California. His mother, Karen Silva, wrote this history. Nick has contributed his own story under the Sharing section on the next page. Nick had his 21st birthday since these articles were submitted for this issue of the Newsletter.

It's been a very long, rough road, but we made it. On May 3, 2000, Nick will be 21 years of age. I haven't written about him since he was very young. Nick does not have classic, intermittent or intermediate MSUD. He has a variant type all his own and has made medical history. The amount of protein he can tolerate is minimal, like in classic MSUD, yet he displays the clinical symptoms of the intermittent type.

He wasn't diagnosed until he was almost 11 months old. After six weeks in the hospital for diagnosis, diet control, and physical therapy he was released to parents who were quite unsure of what life would hold for them and their baby. It was scary. We had entered a world which we had never thought much about.

We were home just two weeks when Nick became ill again. He was hospitalized for several days until his fever came down. So it went for the next five years - so many hospitalizations we lost count. Nick, on several occasions, had hallucinations, convulsions and lapsed into a coma. I remember him vomiting blood once. I always stayed with him night and day, leaving only for short walks to stretch my legs.

I realized early on that the local doctors and nurses really didn't know how to handle this very rare disease, and I had to become the onsite expert. Oh, they did their job, administering medicine and IVs, but beside that, they really didn't know what to do. Several times serious mistakes were made by doctors and nurses. It's scary when you can tell the doctor is panicking. Once a doctor became angry because I called another physician for a second opinion. I told him he didn't get my trust simply because he was a doctor; he had to earn it.

We live 150 miles from the University of California, San Francisco (UCSF) where Nick's metabolic specialist practices, but we couldn't always get there. Most of the time Nick needed immediate care, and it was easier to handle the hospitalization locally.

Nick was sick often when he was little, even when we were on vacation. We went to Minnesota for a week and spent half of it in the hospital there. Nick had caught a flu bug. The staff was great though and made it easy for us to be there. Since then, we've handled most illnesses at home with only a few hospitalizations. Nick drinks apple juice when he is sick and I think it is a major factor in his avoiding hospitalization in the last 6 years. When he was small we used Pedialyte, but as Nick got older, he didn't like the taste of it.

At 5 years of age, Nick went off to kindergarten. While other mothers cried because their babies were leaving home, I celebrated because my son had made it that far.

Nick graduated from high school and attends the local junior college. He also works part time in a department store. In some ways Nick is like the average person. He could walk by you on the street, and he would just blend in with everyone else. Yet if you took the time to look further, you would see the frustration he deals with daily.

Don't get me wrong, you would also see a wonderful, compassionate person. Nick doesn't share his feelings with many people, and it isn't easy for him to talk even to me about how he feels. He sees other people understanding situations and gets frustrated, because he doesn't comprehend what is going on. He wonders why he can't do the things others his age can.

This disease and the limitations that come with it are not too difficult to accept as a child.But as Nick enters adulthood, he understands more fully the limitations MSUD places on his life. He doesn't let many people get close to him. He doesn't like to let them know about MSUD. I think that when he is more comfortable with the disease he will be happier and let more people get close.

Nick doesn't like eating when he is not necessarily hungry and doesn't like going to bed early just to make sure he gets plenty of rest. He doesn't like the headaches or the constant tiredness. Most of all he doesn't like his Mom constantly reminding him of all these things.

In many ways, we are lucky that we live in California. While Nick was growing up he always had private medical insurance.He will no longer have private insurance when he quits school. Along with this, he had California Children's Services (CCS) which acted as his secondary insurance plan. He will no longer have CCS after he turns 21.

Luckily, California has a plan called the Genetically Handicapped Persons Plan (GHPP), and we have begun the necessary paper work to apply for it. Unfortunately, it does not cover anything unrelated to MSUD, such as dental work, vision problems or injuries like a broken bone. We have also started the process of applying for Social Security for Nick. I don't feel he is going to be able to work full time and earn enough to support himself.

This lack of medical coverage greatly concerns me. If he does qualify for Social Security, he will also be able to get Medi-Cal. We are currently checking into some counseling to help Nick in the transition from a child with MSUD to an adult with MSUD.

I have many things to be thankful for. Having a child with a medical problem makes us so much more aware of our immortality. It has helped me to see that there may not be a tomorrow. Be the person and parent you want to be now. Don't wait. I pray a lot. When things are out of our control, all we have left is faith.That keeps me going. Until the next time . . . .
- Karen Silva

FAMILY HISTORIES - THE STORY OF NOA

Details

Written by Joyce Brubacher

Published: 20 July 2009

Noa is the daughter of Yaron and Gally Peled from Israel. The Tel Avivi University first contacted me for information on MSUD, and then referred the grandmother of Noa to me. My first contact with the Peled family was on June 16, 1999.

Our Noa was born on March 31, 1999. She had all the classic symptoms that other families have experienced, and was diagnosed with MSUD on her 10th day of life while in the hospital. The diagnosis was determined very quickly (although it seemed like ages those days), thanks to the professional staff in the hospital.

Life at the beginning (first 3 months) was a bit difficult - getting used to the idea, the stress in the hospital, and it seemed like a very big challenge to balance her amino acids. In addition, it wasn't easy to make her eat, and she vomited a lot.

In Israel there are about 15 children with the disease, therefore, there are very few doctors who have a specialty in MSUD. We learned how to treat her from our doctor in Israel with the help of doctors and nutritionists from the U.S., London and Paris.

We owe a special thanks to Joyce, who supported us from the beginning of our story. She immediately answered every e-mail we sent her, and we learned and gathered a lot of

strength from her. The Newsletter also really helped us to gain the confidence that we were doing all we could, and were being kept up to date.

After 3 months, it got better. Noa stopped vomiting and her development went very well. Noa is now 10 months old and life is normal. Sometimes we forget she has a problem. She is an amazing girl - laughing constantly, never crying and keeping her self busy all the time. And she is s-o-o-o pretty.

The only problem we are facing at the moment is her appetite. Noa doesn't like to eat! If it was for her to decide, she would stop taking her bottle after 5 minutes. So we make many efforts, including singing and playing with her while eating, to get sufficient protein and calories into her.

We are also trying to give her some fruits, vegetables, potatoes and pasta, but we can hardly say that she is 'jumping' for it. We hope it won't be a bigger problem in the future, and that she will eat enough.

It took us some time to realize that the appetite problem is a symptom of unbalanced amino acids. Now we know for sure that it has a connection - when her amino acids are not balanced she refuses to eat at all. But neither is she a big eater when they are balanced.

Like other families, we still have a fear of illness. Till now Noa hasn't been sick, and we are afraid of the first illness, which eventually will come. We would say to each other that all children experience problems, and so does Noa.We especially want her to grow up like every other child (though she is especially beautiful and cute) knowing she has a problem and not a "disease."

We want families to know that we live happily and we would like to help Joyce to persuade others to write for the Newsletter. The Newsletter is a main source for learning and help for our children. To you Joyce - continue in your mission, your work is very important to us.

- The Peled Family

SHARING - MSUD: NICK'S STORY

Details

Written by Joyce Brubacher

Published: 20 July 2009

This candid look at living with MSUD was written by Nick Lovrin. See his mother's account of his life under Family Histories, "Nick at 21."

As a child, I thought that maple syrup urine disease meant that my urine smelled like maple syrup.I really didn't understand why it would smell that way. I have had MSUD since birth. I am one of the lucky ones healthy enough to survive under the conditions of this disease.It affects my physical ability as well as my mental ability.

Let's start with the mental aspect first. Mentally I am pretty smart when it comes to material that I am interested in, but the other stuff is hard for me to learn. It isn't that I can't learn the material, it just takes me a little bit longer to comprehend the basics; then there is no stopping me. Even if I don't do that same thing for a while, I can go back and still have the knowledge and ability to do it. I can succeed at it just like the first time.

Let's take it one more step. I wanted to be a professional basketball player when I was younger. I thought I would make it, but come to find out, I didn't have the height, and I wasn't good enough to play it for a living. So all I do now is play for fun, and I like it.

Then a few years ago, I took up drums for a hobby. For a while, I thought I wanted to do this as a profession. I soon realized, after a little talk and some guidance, that I wasn't cut out to be a drummer. It takes a lot of leg and arm power, which requires a lot of energy to perform during a long concert. I knew I was having a hard time with the drums, because I would be on count (drummer talk) for the first measure, and then I would either increase or decrease the speed making the song sound funny and wrong. It is hard for me to concentrate on one aspect so that it at least sounds right.

This mental block of mine goes back as far as I can remember, and it hurt me a lot in high school.If I didn't have MSUD, I could have pushed myself harder and gotten straight A's, but school was hard for me.I worked hard to do my best, but got mostly C's and D's and only occasionally B's and A's. I guess you could say that I am a little slower than people my own age, and I'm mad about it. All I can say is I'm going to work hard to increase my intelligence to where I can function as normal as possible.

Now about my physical ability, the amount of energy in my body is quickly used up, and I am often tired. If I don't eat or drink my milk (MSUD diet powder mixed in a blender), then I need to have something that has a lot of calories. It will make me feel okay until I get my formula. I can't eat a lot of foods that other people eat, but some of the foods they eat look totally disgusting. If I don't eat right, I could have to go to the hospital where they do a lot of tests and give me shots and an IV. Being stuck with this disorder, for what might be my whole life, I have to work hard to keep myself at my full potential and keep my health in top shape. I have had my slip-ups in the past and have paid dearly for them.

I am upset a lot because I see people, like musicians, doing so much more than me and having fun doing what they do. I wish I could do that, but I have limitations and have to accept them and enjoy the hobbies I like and can do.

I mentioned the drums earlier. I still play them, but I am not as serious about them anymore. I took up singing, which brought childhood memories of when I was ten years of age. I got four of my friends together to form a group that performed for our day camp.We imitated "The New Kids On The Block," because they were what everyone my age listened to. I had fun performing on stage then, and to this day I want to be on stage as a musician more than ever.

I have a lot of obstacles in my way, but I'm willing to overcome them with all my might.MSUD is one of the obstacles, but instead of overcoming it, I will work with it and hope to increase my mental and physical abilities.

Part of the MSUD phenomenon that I have to work with is that I tire quickly. Driving is hard on me.When driving from work to where I live, which is only twenty to thirty minutes each way, I get tired and have to pull over for a nap or just relax a bit. It's just that after a hard day, my body needs to relax, and that is the only way I can make it home. I work in the shoe department of a store part time and attend school two days a week.

School is hard now (it never was easy), but classes are getting harder as I proceed with college.I'm taking two classes at this time, English 80 and Speech 212. English is hard and very boring. Speech, on the other hand, is a blast. The teacher, who is a bit on the weird side, is trying to make us learn more about our inner-self; it is called Interpersonal Communications.

I have another class that starts soon and I'm really anxious for it. It is Golf. I love the sport. When I was younger, and my dad was alive, we would play it. I remember when he would be doing something in the garage, and I got his putter out and started to putt little rocks on the concrete; he got so mad at me. The memory makes me laugh.

MSUD will affect almost anything I do in life, and I have to work around my disorder and make it work for me and not against me. Thanks for reading this little piece I have written.

- Nicholas Lovrin

SHARING - JORDAN'S SURGERY

Details

Written by Joyce Brubacher

Published: 20 July 2009

With Jordan, it is never dull around the Ernest Groff household. His mother, Glenda, our Food News Editor, has shared several of Jordan's experiences, including his very serious illness (in the Dec. '93 issue). In the Fall/Winter 1999/2000 issue, she wrote about their experience with Ritalin.This time Jordan had surgery.

In January, when the last issue of the Newsletter came, I read the article I had written about Jordan. While I was reading, Jordan was lying on the sofa recovering from his last hospital stay. It all started on the 5th of January when I received a call from his teacher at school. "Jordan is not feeling well, could you come get him?"

I don't like calls like that, because you never know what you will find at school. I picked him up and came home. He showed me where his stomach hurt. I called the Clinic and got an appointment to see Dr. Morton. In the back of my mind I thought about his appendix, because Ernie and I both had ours out since we are married.

Dr. Morton was sure it was his appendix and sent us to the emergency room. The surgeon did not think Jordan had enough symptoms to diagnose him with appendicitis, so we went home again.

The next morning I took Jordan for a CT scan. Dr. Morton looked at the scan right away. He talked to the surgeon and they decided to remove his appendix. By 6:30 that evening, Jordan had his appendix removed, but the surgeon was not certain that was the problem.

Immediately after surgery Jordan was started on TPN (Total Parenteral Nutrition) and was on the remainder of the night. His leucine level on the 5th (when he was first checked) was 8 mg/dl and had dropped to 4 mg/dl the day after surgery (two days later).

After lunch on Friday the 7th, when Jordan was drinking and eating again, we were allowed to bring him home. He was in the hospital less than 24 hours. It was good to be home again, because we had a two year old at home with the chicken pox.

Jordan recuperated quickly and only spent six days home from school. It was interesting to hear that the teacher couldn't believe the difference in Jordan when he came back. We wonder if he had been having pain, but didn't want to tell us, because he was NOT going to the hospital. He was rather upset when he had to go directly to the hospital after his CT scan, because I had promised him French fries after the scan. Needless to say, he got his French fries later.

When the pathology results came back, we learned that Jordan definitely had acute appendicitis.We were glad we did the surgery when we did, so he did not get critically ill.

Jordan is back to his energetic self again. I know it took his mother and father a lot longer to recover when we had the same surgery. I guess our age must have been showing!

- Glenda Groff

SHARING - ALL IS NOT WELL IN BRAZIL & IN OTHER COUNTRIES

Details

Written by Joyce Brubacher

Published: 20 July 2009

Joyce Brubacher – Editor

I received a fax from Brazil on March 7 of this year. The urgent message was from Joao Carlos de Oliveira Mello, the father of a recently diagnosed infant with MSUD. His son, one month old, was undergoing dialysis in intensive care. He read on our Web site about TPN (Total Parenteral Nutrition) for MSUD, an IV solution available without leucine, isoleucine and valine that provides total nutrition via IV. It is now the recommended treatment for quickly lowering the branched chain amino acids (BCAAs). The baby was drinking the MSUD formula from Mead Johnson, but he wanted the TPN which is more effective in reducing the high levels of the BCAAs. He wanted to know how to get it from the states as quickly as possible.

Joao and I continued communicating by email. I sent information so his doctor could contact other experienced doctors both in the States and in Brazil. I also sent the information about where to get the BCAAfree TPN. On the 8th, Joao reported the baby was doing better each day.They were praying that their baby would soon be in good health again. They had followed instructions to get the IV solution.

Two weeks later I realized I had not heard from him since the 8th. I sent an email asking how the baby was doing. His reply shocked us. The baby had died the week before. Joao said his son had died of complications from the MSUD. The MSUD TPN solution arrived too late because, as Joao says, "The customs and bureaucracy here is unbelievable."

How very, very sad.Joao and his wife were married 12 years, and this was their first child. Although the family had available resources to transfer the child to the U.S. for treatment, the health of the baby was too poor to permit transport.

I asked Joao if I could tell about their experience in order to expose the plight of families in Brazil and other countries. He mentioned some of the medical problems they faced in Brazil. Only one laboratory in the country could provide BCAA levels in a short enough time to be useful for monitoring. And this was at a university far from their city.The medical food (formula) and TPN solutions have to be imported. In their situation, these were delayed in customs, and an extra charge of 88% was applied for the TPN (which is already very expensive).

Brazil screens only for PKU as is the case in many countries. Statistics in countries such as Brazil are not reliable because many die without a correct diagnosis.

We know there are some success stories from other countries, such as the stories from the Netherlands and Israel in the *Family Histories* section of this issue.The sad stories, however, seem so devastating and so unnecessary.

Another father in Brazil told me the only medical center in a time of crisis is seven hours away by air for his child. A young father of an MSUD child in South Korea made contacts here in the States in order to get DNPH to monitor his daughter. Blood tests for monitoring levels in that country require so much blood his daughter gets anemic each time. He also

told me of hearing of a baby in Chun An, Korea in February 1999.That child was not diagnosed until 2 months of age and died of the cerebral edema. The story in South Korea is much the same as in Brazil - a lack of metabolic centers and very little is known about MSUD.

Following is some information on the situation in the Philippines as related by an Aunt of a child with MSUD. In January 1998 her niece was hospitalized at 9 days of age with problems breathing and moving her bowels. After 5 days she was much worse and was put on a respirator. A neurologist suggested doing a newborn screening test.In the Philippines there is only one laboratory to do these tests.

The infant was diagnosed with MSUD after two weeks in intensive care. She was discharged after two months in the hospital. At two years of age her behavior is like that of a 2 month old. A fax I received in December 1999 pleaded for help for funds to buy some formula.The child was getting 2 cans free a month from Mead Johnson and that was to end at the end of the year.

It is heart-wrenching to know there are many children with MSUD dying in other countries, or who may be diagnosed soon enough to survive, but have no funding for formula or good follow-up care. This is true in Chile, where most children with MSUD are stunted in growth and development, because the families cannot afford formula.

We have received reports from doctors and families of children receiving improved treatment because of what they have learned on our Web site. We hope to improve the site in the near future and add an index.The least we can do is share knowledge with one another through our Symposiums and Web site. The Lord will add His blessings.