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MSUD AND PREGNANCY

Details

Written by Sandy van Calcar

Published: 20 July 2009

In the December '90 issue of the Newsletter we congratulated Sue Ann McNight for making medical history. She gave birth to a healthy 5 pound, 12 ounce baby girl on November 17, 1990. Sue Ann has classic MSUD. Following is an account of her second pregnancy as reported by her metabolic nutritionist, Sandy van Calcar from the University of Wisconsin Biochemical Genetics Clinic in Madison, Wisconsin. Sue Ann shares her own account of her struggle with Guillain-Barré Syndrome and her pregnancy and delivery of her son in the SHARING section of this issue.

As many of you know, Sue Ann McKnight is one of the oldest individuals with MSUD who has been treated since infancy. She delivered a healthy girl, Amanda, about 6 years ago and the outcome from that pregnancy was published (see references). Amanda is now in kinderarten and is doing great! Sue Ann delivered a baby boy, Blake, in March of this year [1997]. I'd like to share some of the details from this pregnancy.

Sue Ann has a higher tolerance for leucine than many individuals with classic MSUD. Because of this, she counts grams of protein rather than leucine and is allowed 29 grams of whole protein from food and whole milk which she adds to her formula. This amount of protein is equivalent to 2000 mg of leucine per day. When Sue Ann gets ill, she rarely has problems because of her MSUD. Since her last pregnancy, she suffered from metabolic acidosis only once when she developed Guillain-Barre Syndrome three years ago. (Guillain-Barré Syndrome is caused by nerve inflammation which results in progressive weakness and paralysis.)

Sue Ann presented to our clinic with her second pregnancy at 5 weeks of gestation. Her leucine level was 191 µmol/l (2.5 mg/dl). To meet her nutrition needs during pregnancy, we increased her protein equivalents from formula by adding MSUD 2 to her usual formula of Ketonex II. She also started a prenatal vitamin and extra calcium. Her leucine prescription was not changed.

During this pregnancy, Sue Ann's weight gain was normal and DNPH remained negative. We followed the same lab monitoring protocol that we developed after her first pregnancy (*see Table 1*). Her free carnitine in plasma decreased below 20 µmol/l at 8 weeks gestation and so carnitine was started at 20 mg/kg. During both the first and second trimester, her leucine prescription changed little (*see Table 2*). An increase of only 200 mg leucine in her diet increased plasma leucine above 200 µmol/l (2.6 mg/dl).

Table 1: Suggested Monitoring Protocol for Pregnant Women with MSUD*

Analyte	**1st & 2nd Trimester**	**3rd Trimester**
Urine ketones	Daily	Daily
DNPH	Weekly in the morning	Daily in the morning
Amino acids (Pl & Ur)	Every 2 weeks	Weekly
Carnitine (Pl & Ur)	Monthly	Every 2weeks
Urine organic acids	Once per trimester	Every 2 weeks
Albumin	Once per trimester	Once per trimester
Hemoglobin & Hematocrit	Once per trimester	Once per trimester
Weight gain	Every two weeks	Weekly

*Assuming no signs of metabolic decompensation are noted. From reference #2

Table 2: Average Plasma amino acids and dietary intake for second pregnancy

	Leucine	**Valine**	**Isoleucine**	**Average leucine intake**
µmol/l	µmol/l	µmol/l	(mg)	
1st Trimester	128	143	65	2000
2nd Trimester	167	206	81	2000
3rd Trimester	288	303	131	2200

Normal values:	leucine	120 +/- 32 µmol/l (1.6 mg/dl)		
	valine	225 +/- 50 µmol/l (2.6 mg/dl)		
isoleucine	57 +/- 20 µmol/l (1.6 mg/dl)			

An ultrasound at 16 weeks of gestation showed that the baby was small but had normal development for his age. We increased her carnitine and protein equivalents from formula to see if this would improve the baby's growth. Another ultrasound at 23 weeks of gestation again showed a small baby, but normal development and growth rate. Sue Ann's metabolic labs remained within normal range.

Ultrasound at 27 weeks of gestation showed that the baby was falling from the 10 percentile for growth and a nonstress test showed decelerations, or slowed blood flow, for the baby. Sue Ann was admitted at that time for further evaluations. Her metabolic labs were again normal. We increased her whole protein intake and carnitine dose. An additional 280 mg leucine in diet increased her plasma leucine to 417 µmol/l (5.6 mg/dl).

During the two week admission, the baby was monitored daily with nonstress tests and biophysical measurements. These measurements were encouraging until 29, weeks of gestation when the tests again showed fetal decelerations with decreased tone and activity. A small pleural effusion and ascites (fluid in lung cavity) were found on ultrasound. Because of these problems, the baby was immediately delivered by C-section. He weighed 584 grams (1 pound 4.6 oz) and his length was 11 inches. Apgar scores were 3 and 7 at 1 and 5 minutes.

The baby initially required mechanical ventilation and had other problems typically associated with prematurity including bronchopulmonary dysplasia, jaundice, anemia and retinopathy. He required a neonatal ICU stay of 83 days. At discharge he weighed 4 pounds and was 16 inches in length.

Fetal growth and Sue Ann's leucine tolerance were much different with this pregnancy than with her first one. During the first pregnancy, her tolerance for leucine was much greater. At the end of the second trimester she was tolerating 3800 mg leucine compared with 2200 mg during the second pregnancy. With the first pregnancy, fetal growth delay was detected much later in pregnancy (32 weeks of gestation) and it was not as severe as with the second baby. When we detected the growth delay with her first pregnancy, we were able to gradually increase her leucine intake without causing elevated plasma leucine levels. By the end of her first pregnancy, she was taking 8600 mg of leucine per day without causing an increase in plasma levels. This was 4600 mg more than her prepregnancy tolerance!

During the first pregnancy, she also needed more carnitine to maintain normal blood levels. Her final carnitine dose was 150 mg/kg for the first pregnancy compared with a dose of 50 mg/kg for the second one. For the first pregnancy, both the increase in protein and carnitine

seemed to improve the baby's growth during the final 6 weeks of pregnancy. With the second pregnancy, the baby's growth did not improve.

After her second delivery, studies of placenta tissue showed multiple small thrombosed vessels (blood clots) which were consistent with chronic ischemia (deficient blood supply). These findings could explain the baby's small growth since he likely was not receiving adequate oxygenation or nutrients. The deficient placental blood flow could also explain why Sue Ann's branch chain amino acid tolerance did not increase during the second pregnancy. Since the fetus was not receiving adequate amino acids via the placenta, Sue Ann's plasma leucine levels increased even with small increases in intake. We do not have any idea whether MSUD had a role in the development of poor placental blood flow. It seems unlikely given Sue Ann's excellent metabolic control, although conceivably, there may have been a period of high levels that we did not know about.

Blake is now 8 months old and despite the pregnancy complications, he continues to catch-up in both growth and development. We are optimistic that he will not have any long term complications from his prematurity. I know that he will have every advantage because of the great care he will receive from his parents, Sue Ann and Loni.

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RESEARCH AND CARRIER TESTING FOR MSUD

Details

Written by Joyce Brubacher

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At Symposium '94 in Columbia, Missouri, Julie Grasela, R.D. used a Q-tip to take samples of cheek cells from volunteers. These samples were used for DNA tests in an effort to see if non-Mennonite persons with MSUD could be carriers of the specific classic type of MSUD found in the Mennonite population. This report was submitted September 30, 1997 for our MSUD Newsletter. It explains the studies being done at the University of Missouri Medical School, Columbia, Missouri.

Drs. Charlotte L. Phillips and Richard E. Hillman of the University of Missouri School of Medicine, Departments of Biochemistry and Child Health have an ongoing research

program investigating the genetic basis of maple syrup urine disease (MSUD). Dr. Phillips is an Assistant Professor, with a Ph.D. in Biochemistry, and a Clinical Molecular Geneticist. Dr. Hillman, who is familiar to many of the MSUD families, is a Professor, Physician, and Metabolic Geneticist. Drs. Hillman and Phillips have put together a research team whose current programs and research studies include:

1. **Genetic testing for the MSUD alteration common to the Mennonite community; providing carrier detection and newborn screening for Mennonite communities.**
   For review, MSUD is an autosomal recessive metabolic disorder due to a defect in the branched-chain a-keto acid dehydrogenase (BCKAD) complex which catalyzes the oxidative decarboxylation of the a-keto acids derived from leucine, isoleucine and valine. Clinically the effects are due to the inability to breakdown protein, specifically certain amino acids (isoleucine, leucine, and valine). Children born with classic MSUD are healthy and appear normal at birth, but within 7 to 10 days they have poor feeding, become lethargic, go into a coma and die if untreated. Treatment is essentially a low protein diet with controlled amounts of isoleucine, leucine, and valine. Infants, whose treatment is begun late, are at risk of neurological damage and mental retardation. It is believed that the earlier infants are identified, the better the prognosis.

   The prevalence of MSUD in the general Caucasian population is 1:225,000. In Old Order Mennonite communities of Pennsylvania, the reported incidence of MSUD is 1:176. In Missouri the incidence of MSUD in the Mennonite communities may even be higher, 1:80. Mennonite MSUD patients were found to be homozygous for a T to A transition resulting in a Tyr to Asn change (Y393N) in the E1a subunit of the decarboxylase of BCKAD. This means that both copies of the E1a gene that were inherited, one from mom and one from dad, contain the Y393N alteration and is the reason these patients have MSUD.

   In response to the request of two Missouri Mennonite communities for carrier testing, Dr. Phillips' laboratory developed a noninvasive, quick and accurate DNA test. The Y393N alteration can be easily detected using DNA isolated from the loose cells that line the inside of the mouth. Mennonites from several communities have utilized this carrier testing. Drs. Phillips and Hillman hope to identify all the families at risk for an infant with MSUD, in order to insure rapid detection and treatment of the newborn. Those families in which both parents are carriers, can now be prepared to have their newborn infants tested immediately after birth. The hope is to identify the MSUD infants (within 36 to 48 hours after birth) and place them on the special diet before they become ill or can suffer any neurological damage. Four Mennonite families have already benefitted from this newborn screening.

2. **To clearly define the specific alterations in the E1a genes which result in maple syrup urine disease.**
   In addition to testing all the Mennonite individuals who requested testing, we have also examined non-Mennonite MSUD patients who wished to be tested. In the non-Mennonite MSUD population (many of which were sampled during

the MSUD symposium in June 1996, Ohio), we determined that approximately 25 percent of the non-Mennonite MSUD patients had the Y393N alteration in at least one of their E1a genes. Two non-Mennonite patients were homozygous for the Y393N alteration (both E1a genes had theY393N alteration, similar to Mennonite MSUD patients). Six patients were compound heterozygotes for the Y393N alteration. Since MSUD is an autosomal recessive disorder (requiring both E1a genes, one from mom and one from dad, to be defective), these six patients have the Y393N alteration in one of their E1a genes, and a different alteration on their other E1a allele.

Determining the other alterations which result in nonfunctional E1a genes is one of Drs. Phillips and Hillman's objectives. Their hope is that knowledge about the alterations which lead to nonfunctional E1a will provide the opportunity to better understand the important structural/functional domains in E1and may lead to better or alternative treatments. An additional benefit to the immediate families is the knowledge of their own cause for MSUD and may permit more informed prenatal and postnatal genetic counseling.

3. **To determine the genetic origin of the Y393N alteration (common to Mennonite MSUD patients) in the non- Mennonite MSUD patients. Do these patients have Mennonite ancestors or is this region of the E1a gene susceptible to becoming defective?**
   All the Mennonite MSUD patients tested were found to be homozygous for the Y393N alteration in the E1a subunit of the decarboxylase of BCKAD. It is believed that all of the Mennonite MSUD patients inherited their Y393N alterations from a common Mennonite ancestor. Dr. Phillips is interested in determining if the Y393N alteration in the non-Mennonite MSUD patients is from this same Mennonite ancestor, or is it an alteration that arose independently, because this part of the E1a gene is more prone to error or change. By using DNA markers and examining the DNA near the Y393N alterations, this question should be answerable.

This work would not be possible without the volunteer time and efforts of the clinical and research staff at the University of Missouri School of Medicine and donations from MSUD families to the Medical Genetics Research Fund.

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FAMILY HISTORY - KRISTA OLSTAD

Details

Written by Todd Olstad

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The Oldstads sent this history in April '97 along with a newspaper clipping of an article describing Krista's care and the way it has affected her family. The article included a large color photo of the family - Todd, Carlene, and their two children, Trevor and Krista. It is encouraging to see the media publicizing MSUD.

We had a daughter, Jessica, who passed away twelve years ago. The doctors had discovered that her diaper smelled sweet - that's how they found out that Jessica had MSUD. But the doctors were unable to figure out what was wrong soon enough to treat her. For this reason our pediatrician, Dr. Bilgi was aware of what to look for when our daughter Krista was born.

Krista Olstad was born at Mercy Hospital in Des Moines, Iowa on January 29, 1996. It was a normal birth - everything went quickly for us. As soon as Krista was born, they started their testing. The day we were to be discharged, Dr. Bilgi called and said one of Jessica's diapers had a sweet odor to it. We put Krista into the ICU at the hospital and started the treatment for MSUD. The following day we got some test results back which indicated MSUD. We transferred Krista to the University of Iowa Hospitals. There Krista was put in the hands of Dr. William Rhead, Judy Miller, and Cheryl Stimsom.

Carlene went with Krista that night. I stayed home to take care of our son, Trevor, and to explain to him what was going on with his sister. Trevor, who is six years old, doesn't have MSUD. He stayed with friends for a few days. We were about two hours away from home.

The next day Carlene and I met with our doctors and others who told us that Krista has MSUD. During the next few days we were told a lot more about the disease. Krista was in intermediate ICU. We both were very nervous the first few days because it's a very big hospital, and we were hoping that Krista wouldn't get worse. Our doctors were great in keeping us informed about her progress.

Everyone was very helpful the whole time we spent there. The nurses in that ICU let Carlene and me help do things so that we could stay busy. Krista was on an IV and also bottle feeding. At times she didn't want anything to do with her bottle.

They had to put an NG tube in Krista and feed her that way most of the time. Carlene and I were able to help feed her and we were also taught how to change her tube which was sometimes hard to do. Judy Miller showed us how to do this and how to check the ketone

level in her urine. We put cotton balls in her diaper and then put the wet cotton in a syringe to squeeze out the urine to collect it for the test. **(Great idea!)**

Cheryl Stimsom showed us how to mix Krista's formula. The first few days at home we had problems with this, but we called Cheryl and Judy about it. They helped us. I had started to mix the formula and did it wrong. Now Carlene mixes it every day. It is better to have one person do the mixing, but I know how in case I have to.

Judy had to teach us how to do heel sticks - we are using Krista's toes now. That blood goes to New York. When we have blood drawn at our hospital, it goes to Utah. It used to go to the University of Iowa. They had to close the lab there which really worried us. They had done our tests right away for us, so we were always getting a quick turn around on results.

Krista was at the University of Iowa hospital for ten days. We were nervous about taking her home. But she has been hospitalized only once in her first year. That was for one week. Krista had a cold and ear infection. Dr. Bilgi had seen her a day before she went into the hospital. We thought the medication she was on hadn't taken effect yet. Krista was sleepy and whining that day. When I checked her ketones that night, her levels were real high. We called Dr. Bilgi right away and he told us to take Krista to the hospital immediately.

She was admitted, started on an IV, and taken off her formula. After a couple of days, she got back on formula. After a few more days, her levels starting coming down. We got real scared when her ketones got that high. We had never had anything but a negative reading on her Keto Stixs.

Krista has been easy to manage with her MSUD. She started walking at about ten months. She is real good in nature - always happy and loves to play with her toys. Krista also loves to give hugs to people and play with her brother and other kids.
Lots of family and friends helped us out when we were at University of Iowa Hospitals. We see Dr. Rhead, Judy Miller, and Cheryl Stimsom at least once a month at the Children's Clinic at University of Iowa, and see Dr. Bilgi when Krista needs a checkup. All these people have made it easy for Carlene and me to keep Krista healthy. We think the world of them for being so caring.

Update: December 1997
Since we sent the history in April, Krista has been in the hospital twice, the last time being the week before Thanksgiving. Both times she had a flu which sent her ketones up. Her three amino acid levels went up a little, too. So we put her in the local hospital on an IV for her dehydration, but each time it was only for a week.

Krista has been really doing well. Her weight is about 25 pounds and height is about 31 inches. Sometimes she doesn't want to eat certain things, like her peas, green beans and some other vegetables. But Krista loves corn, bananas and popsicles. We got some low protein recipe books, so we're able to start making more foods for her. Krista's formula is a mixture of Ketonex and Isoleucine and a little 2% milk. My wife sometimes makes some of it into a paste which Krista takes along with water and other fluids.

We do checkups at the University of Iowa every couple months now. They say Krista is doing really well. She is doing what every 21 month old is doing. She is learning a few new words every day with the help of her older brother who is eight.

We take Krista to a local lab to have blood drawn every four weeks to test her three amino acid levels. This blood is sent to a lab in Texas. In the weeks between those draws, we draw blood at home using filter paper and send it to New York to check her leucine level. This all helps us regulate Krista's Diet.

Krista does well in playing with other children at day care. We have a very good day care. They have had Krista since birth and really watch her. If they sense something is wrong, they call one of us right away. We have a lot of good people that take good care of Krista.

FOOD NEWS

Details

Written by Joyce Brubacher

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New, Wonderful Low Protein Cheese
It looks like real cheese and the taste isn't far behind. It is low protein mozzarella and cheddar cheese from Ener-G Foods. A great new product which comes in 3 lb. blocks. The cheddar cheese has 1 gm protein per ounce and 80 mg leucine. The mozzarella cheese has less than 1 gm protein and 63.3 mg leucine per ounce. It keeps in the refrigerator a little longer than regular cheese and can be frozen.

Use this low protein cheese just as you would regular cheese. It does not melt, just softens, but is very attractive and tasty in salads, grated on pizza, sliced for sandwiches, and used in mountain pies. Check Ener-G Foods, Inc. under Resources.

Cheese Flavored Sauce Mix
Dietary Specialities markets a delicious powdered cheese sauce mix. Just add water and heat for a rich, cheese flavored sauce. The 5.64 oz. jar makes 4 cups of sauce. It can also be sprinkled dry on top of casseroles or low protein crackers or mixed with low protein pasta. They also market the Crackertoast and the lasagna noodles in the preceding recipe. Check Dietary Specialities, Inc. under Resources

Loprofin Foods
SHS North America now offers a variety of tasty low protein products - baking mixes, canned bread, pastas, crackers and wafers. They also sell an International PKU Recipe Book with 120 pages of recipes. For more information send to SHS North America. See under Resources.

SHARING - SUE ANN HAS BEEN SHARING, EDITOR'S NOTES

Details

Written by Joyce Brubacher

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The first issue of the MSUD Newsletter was dated February '83. The second issue, November '83, included an article sent to us by Sue Ann Fredericks from Wisconsin. She was 17 and had written the paper for her senior psychology class. First, she described MSUD and then gave her personal history. A brief summary of that history will give you some background for her story which follows.

Three years prior to her birth, her parents had a son who died undiagnosed at four months. When Sue Ann was born, her mother noticed the same sweet smell and similar symptoms. Sue Ann was in a coma when she was diagnosed at 11 days.

She spent most of her first three years in the hospital, facing death twice. She refused to drink her formula, so her mother mixed it with baby rice cereal until she was five. Sue Ann writes that her worst years were between the ages of six and eleven. She was in the hospital at least three times each year with summer being the worst time. She would feel dizzy and lightheaded during recess, stagger, and talk without making sense. She would see "things," not recognize people, and then start vomiting. She needed to be in air-conditioning.

Sue Ann's early battles affected her coordination and balance so that she could not ride a bicycle. She did well in school, graduating from high school in '83. She received a diploma in Child Care Services from a technical college in '84 and has been steadily employed since. We announced her May 1988 marriage to Lon McKnight in our June '89 Newsletter.

In her history, she made this interesting observation, "My body chemistry may not be able to handle all the changes of pregnancy, so I may not be able to have children." She proved herself wrong with the birth of her first child, a healthy 5 lb. 4 oz. baby girl born November 17, 1990. She shared an account of her pregnancy and delivery of Amanda in our December '90 issue.

I think it is it is very commendable that Sue Ann is again willing to share by relating the following details of her illness in '94, her pregnancy in '97, and the birth of their second child. (For medical details see page one of this Newsletter.) Adversity can be a refining process and I trust these trying experiences will make Lon and Sue Ann stronger persons.

SHARING - SUE ANN MCKNIGHT, MY STORY

Details

Written by Sue Ann McKnight

Published: 20 July 2009

My name is Sue Ann McKnight. I have had Classic MSUD all my life. I was diagnosed at eleven days old by Dr. Henry Waisman in Madison, WI. I am now thirty-two years old.

At the end of January 1994, I got the flu. I felt run down; I was sneezing, coughing - the works. Then my fingertips and calves went numb. They felt like they were asleep, only they never woke up. It was not that much of a problem until my fingers starting slipping off the keyboard of my computer at work. I went to see my internist, Dr. Frank Polyak on February 2. He didn't know what was causing the numbness. He sent me to a neurologist, Dr. Murphy. I was scared. I work at an insurance company. I know what a neurologist does.

Dr. Murphy did a variety of tests (made me squeeze a ball, push him as hard as I could, etc.). He tested my reflexes with a rubber hammer. My reflexes were gone. Nothing happened when the hammer struck various parts of my body (knee, ankle, etc.). Dr. Murphy diagnosed me with Guillain-Barré Syndrome (GBS). I had heard of GBS, but didn't know what it was.

GBS is a debilitating disease that attacks the nerves. It is caused by an antibody from an infection (such as the flu). Usually, the body produces antibodies to fight the infection. In the case of GBS, one of the antibodies turns against the system. Around every nerve in the human body is a cushion, so when you bump something, it doesn't hurt that much. With GBS, the bad antibody destroys the cushion around the nerves, making them highly sensitive and paralyzing them. The only cure is strong antibodies and time.

Dr. Murphy put me in the hospital to watch my condition. If GBS gets in the chest, it can kill by paralyzing the lungs. I was in the hospital four days. The GBS was in my arms and legs, but I could still walk, feed myself, etc. I felt fine, just numb. When I was discharged, Dr. Murphy said I could go home but not back to work. Another thing GBS does to a body is wear it out. The littlest thing can tire the body tremendously. He didn't want me to overexert myself.

So for two weeks, I stayed home, resting as the doctor ordered. Then one Monday, I woke up and was so dizzy that I couldn't walk down the hall without hitting the walls on either side. My husband, Lon, told me to call the doctor as soon as possible. I called Dr. Murphy's office at 8:30 when it opened. He told me to go to the Emergency Room as quickly as I

could. I called Lon. It took him ten minutes to get home. I couldn't stand up. Lon helped me into the car and took me to the Meriter-Park Hospital Emergency Room.

The numbness was spreading fast. It went to my face. I had a hard time talking. My facial muscles were numb. I stayed in the ER for a few hours, and then I was put in my own hospital room. By then I couldn't move. My hands locked up into fists. I had IVs to prevent dehydration. They gave me a very powerful antibiotic called gamma globulin intravenously. Lon stayed with me until visiting hours were over. Then he picked up our then three year old daughter, Amanda, from my parents' house.

The only things I could move and control were my head and neck. So the nurse pinned my call button to the collar of my hospital gown in case I needed help. I remember lying in bed. Suddenly I started breathing real fast and I couldn't slow it down. I frantically hit my call button with my chin. The next thing I knew, it was the morning after. Lon and my mom were in my room. I had a tube down my throat and my glasses were missing. I didn't find out what happened that night until two months later. Lon waited to tell me until I was stronger and able to handle it.

He had gotten to bed about 10:30 p.m. and just fallen asleep when the phone rang. The doctor told him to get back to the hospital immediately. He took Amanda back to my parents, and he and my mother rushed to the hospital. Lon had been told to go to the ER because the main doors were locked. A security guard met them at the entrance and took them up to ICU. A priest met them in the waiting room. He told them I had gone into Code Blue and the doctors were working on me. GBS had gotten into my lungs.

I was on a respirator for a day and a half. I was in ICU for two days then moved to a private room. Then the pain set in. It was unbearable. The only way to describe the pain is - take the pain of when your leg wakes up after it falls asleep and multiply it one hundred times. My nerves were so sensitive. I couldn't stand to be touched, especially my hands and feet. GBS in the extremities (fingers and toes) takes longer to rehabilitate because they're farthest away from the body. For two weeks, I was miserable. I was paralyzed and in pain. No one knew what I was going through. No one could really comfort me because of the hypersensitive nerves and pain. I became very depressed.

I was transferred to the Rehab Floor where they help stroke victims, accident victims, anyone who needs to learn how to do things all over again because of a medical condition. I had four sessions of therapy, Monday through Friday, and two sessions on Saturday morning. I had physical therapy (PT) and occupational therapy (OT) - one session of each in the morning and one of each in the afternoon plus one of each Saturday morning. PT was for my lower body so I could learn how to walk again. OT was for my upper body so I could get back to my job and work with a computer again. In OT, I sorted beads, put puzzles together, and worked on the computer to help my hands get back to normal. In PT, I learned how to walk all over again with rigorous daily exercises and routines.

I went home the last week of April 1994. I went home with a rolling walker that had two wheels in front. I continued my physical therapy at home for a month. I could walk but I still had some numbness in my feet. I went back to work part-time in July and full time in August.

Two years later in September 1996, I took a home pregnancy test. It was positive. I had an appointment with my family practitioner, Dr. Russ Hermus, to check why my feet were hurting (I had a burning sensation when I walked). He confirmed the pregnancy and estimated my due date as April 4, 1997. I had an ultrasound later that day. Everything was normal. We told our families and I consulted with my dietician, Sandy van Calcar. She changed my formula recipe to regulate my diet. I had to keep close track of the daily amount of protein I consumed. I had to have a certain amount of protein every day and take Carnitor and MSUD-2 pills.

My due date turned out to be May 20, 1997. Dr. Hermus set us up with Dr. Susan Davidson, a perinatologist (high risk pregnancy doctor) at St. Mary's hospital. She took over my case.

In January 1997, I broke my left foot and had to be in a wheelchair for six weeks. The second week of February we had an appointment with Dr. Davidson for an ultrasound. She was worried about the results and wanted us to come back the next day to be monitored. They hooked me up to a monitor to check the baby's movement and the way the fluid moved through the umbilical cord. It also recorded breathing and heartbeat. There were times during the monitoring that it seemed as though the baby was crimping the cord and not enough fluids and/or oxygen were getting to the baby. Dr. Davidson didn't like the results so she admitted me to the hospital.

The baby wasn't growing as he should. So they monitored me for twenty-four hours straight, but since the problem didn't have any pattern to it, and was very sporadic, they decided to do it once in the morning and once at night. I had ultrasounds daily. They checked four different things on the ultrasound - the amount of amniotic fluid, two types of movements, and breathing. You rate two points for each. The best you can get is eight out of eight. Most of the time, I got a six out of eight. A couple of times, I did have eight out of eight. But still, for some reason, the baby wasn't growing the way it should.

Four days after I was admitted, I had slight contractions and the monitor showed a pattern to the cord problem. I was rushed to labor and delivery. An IV was put in. They were going to prep me for a cesarean section. Dr. Davidson discussed the options with Lon and me. My contractions had stopped and the cord problem lost its pattern. I was at twenty-eight weeks in my pregnancy. I could either have a C-section that night or hold off until my thirty-second week to have the C-section when the baby would be a little further along. We chose to wait until the thirty-second week. I began taking steroid shots. Because the baby would be born prematurely, his lungs would be underdeveloped and the shots would help the baby.

On March 6, 1997, I went for my daily ultrasound. The technician saw something wrong. He got Dr. Davidson right away. On the ultrasound, the baby was in trouble. One lung was filling up with fluid. I started to cry. I called Lon to get to the hospital. He was there in eight minutes. I was prepped for a C-section. An IV was put in. I was given a spinal epidural which made me numb from the abdomen on down. I was awake through it all. Lon sat next to me and held my hand. It took thirty to forty-five minutes (from prep time) to get the baby out and another forty-five minutes sewing me up. Blake Andrew McKnight was born at 2:44 p.m. He weighed one pound, four and one half ounces and at eleven and a half inches was a little bigger than his daddy's hand.

He was taken to the Neonatal Intensive Care Unit (NICU). He had an IV and was put on a ventilator. He was hooked up to different monitors - one checked his heart rate, one checked his breathing; his pulse, everything was watched. I got to see him about three hours after he was born. They wheeled me, bed and all, into the NICU. I never saw anything so small - I was amazed. He was perfectly formed, just small. They fed him through an IV. He had to have blood transfusions, because he didn't have a lot of blood in him, since he was premature.

He stayed on the ventilator until March 19. On March 26, his oxygen levels had fallen, so his doctor, Dr. Dorothy Ritter, decided to give him oxygen through a nasal tube. He showed what a fighter he is, because he pulled this tube out frequently. On March 27, a feeding tube was put in to replace the IV. We went to visit him every night. Amanda went with us. On April 7, he finally got rid of the nasal oxygen. The doctors discovered Blake had a double hernia, which is common in premature babies. This had to be operated on before he went home. I got to feed him through a syringe on April 13, 1997. He was given steroids to help his lungs. By April 23, he was being bottle fed. By May 7, he was a little over three pounds. On May 19, he had the hernia surgery. It went well. Blake finally got to come home on May 28, two and a half months after he was born.

He is now seven and a half months old and doing very well. He's cooing and making noises, drooling, rolling over, eating cereal and fruit, etc. Amanda, now seven years old, is in first grade and loves being a big sister.

SHARING - KEITH MARTIN GRADUATES

Details

Written by Joyce Brubacher

Published: 20 July 2009

Keith received the Best Student Award when he graduated from the Brownstown Career and Technology Center (BCTC) on May 29, 1997. He attended BCTC half days for two years while still enrolled at Garden Spot High School. He was in the Basic Occupational Skills Class the first year where he learned a variety of job skills. The second year he was mainstreamed into classes such as construction, masonry, painting and decorating. On June 9, 1997, he graduated from high school.

Presently Keith is working full time at the Shady Maple Cafeteria as a busboy and dishwasher. After driving with a permit for 7 months, he got his driver's license on Dec. 4. Three days before, on Dec. 1, he shot a nine point buck, his first deer! Quite an accomplishment for a 16 year old hunter and what an exciting week. Congratulations Keith, and our best wishes for a successful future as a worker and hunter.

SHARING - EMILY'S STORY, AN ADULT WITH MSUD

Details

Written by Emily

Published: 20 July 2009

When I met Cindy Blau at our Symposium '96, I assumed she was the oldest living person with MSUD. I had read about her in medical articles, where she was described as the first person to be treated for MSUD. She was born in 1959.
Then Emily contacted our organization earlier this year. When I saw her Newsletter subscription form, I was astonished to see that she was 39 years old. It seemed incredible that there was someone with MSUD who was older than Cindy. I just had to call her, and was immediately impressed with her soft, articulate voice and her interest in our organization. Emily and I have been getting acquainted through e-mail ever since.

Emily was quite willing to share her story and some of her art work with us. When I read her story, it deeply affected me. Her childhood was so sad and she endured much suffering. It is amazing and a tribute to her fortitude that Emily maintained her own diet restrictions without much help for so long. I look forward to meeting her at our next Symposium.

Maple Syrup Urine Disease was identified as a disorder in 1954. I was born in 1957, on August 24 - the third child of four. Both my younger brother and I were born with MSUD.

My two older siblings had normal metabolism. I have almost no visual or audio memories of my childhood before the age of 6, only emotional/ feeling ones. For example, drinking filtered apple juice always causes me to cry - I suspect that this is because of an unconscious association I formed between being fed filtered apple juice and being hospitalized frequently as a baby. I have never sat down and written about my life in the context of living with MSUD - I hardly know where to begin, what to tell you or what to skip.

My parents had me when they were in their late twenties. They had met in medical school. My father became a doctor. My mother completed her third year of medical school but then dropped out when she had her first child, never to return and complete her M.D. My father battered my mother for almost the entire 13 years of their marriage. He was also often very violent towards his children. My parents were in the midst of their divorce when my little brother died from brain edema due to the MSUD. He was four years old when he died, and I was six. His death resulted from a poorly done tonsillectomy - he was not stitched up properly and so blood kept leaking into his stomach. This heavy influx of protein into his body caused him to lapse into a coma - he died after being on life support for approximately 72 hours. My parents never told me that David had died from the MSUD. It wasn't until I

was about 30 that I found this out through reading a medical journal article which discussed myself, my little brother, and several other kids with MSUD.

I was a healthy baby, showing no signs of MSUD, for about the first six months of my life. My mother always attributed this to the fact that she breast fed me for those first six months. It wasn't until my mother took me off of breast milk and started feeding me cow's milk that I became very ill. Doctors were absolutely baffled as to what could possibly be causing me to be so sick. The slightest infection or cold would cause me to lapse into a coma for several days on end. I was constantly being sent off to hospitals, sometimes as far away as 100 miles from home.

It wasn't until I was about 1, years old that doctors finally diagnosed me as having MSUD. My parents had noticed a sweet smell to my urine and ear wax, and this proved to be an important clue. Many medical procedures were performed on me in attempts to come up with a diagnosis, as well as to keep me alive. As a baby, I had holes drilled into my skull because doctors thought I might be passing out all the time due to a brain tumor - the holes were meant to relieve pressure on the brain. In my later childhood years, my family often teased me about having had holes drilled in my head. Because of how they joked about it, and how unbelievable it sounded to me, I had always assumed it couldn't possibly be true. However, when I was in my thirties, I was getting my hair cut one day and my hairdresser mentioned to me that I had two long parallel scars on the back of my head. The first words out of my mouth were, "My god, it's true."

As a young child I remember feeling extremely ashamed of what I ate. I have vivid memories of lunch time in grade school. I would sit with all the other children at those big cafeteria tables. I brought my lunch from home and it was the same every day - two slices of white bread with mayonnaise on it and a separate plastic baggy with head lettuce in it. I would have to assemble the sandwich myself at lunch time. I was so ashamed of eating differently from the other kids that I would always assemble my sandwich under the table so that no one would see what I was eating. I knew that other people put lots more things on their sandwiches and I felt ashamed that mine was so lacking. I remember always feeling afraid and anxious that someday somebody would see me putting my sandwich together under the table, notice what I was eating, and treat me mean because of it.

I have memories from when I was about six years old of staggering down the hallways at home, banging into walls and feeling very confused. In retrospect, I understand that I was suffering from ataxia due to the MSUD. To my recollection, my parents never took me to the hospital or got me medical help during those times.

My father remarried when I was about seven years old. My stepmother cooked meals for the family every night, but never made it a point to prepare foods that I could eat. I remember, night after night, walking into the kitchen very quietly after all the other family members had sat down in the dining room to begin their supper meal. I would look at the food on the kitchen counter to see if there was anything I could eat. (To my recollection, my parents never tried to help me or teach me what foods I could or could not eat - except I knew that I should not eat meat. I instinctively gravitated towards starches, high carbohydrate foods, and low protein foods). From the food on the kitchen counter, prepared by my stepmother, I would usually not find anything I could eat. As a child, I remember thinking to myself at those times, "If only I would just eat normal like everybody else there

wouldn't be a problem here - it's only because I'm choosing to not eat what everybody else is eating that there's no food for me to eat at dinner."

There was one thing that I knew how to cook at that age (because I had watched my stepmother make it sometimes) and that was Potato Buds. So I would reach up into the cabinets and get the box of Potato Buds to make my dinner. I would do this as quietly as possible for fear of offending my stepmother. As a child I thought she would interpret my making my own dinner as an insult to her. I was afraid she would think that the reason I wasn't eating her food was because I thought she was a bad cook. This dinner ritual played out every single night from about the age of 7 to 19 when I moved out of my stepmother's house (my father was dead by then - he committed suicide, secondary to terminal cancer, when I was 15).

Occasionally, my stepmother's relatives came over for dinner and they would usually ask about me. They would ask why was I eating so little food, and why I wasn't filling my plate with all the foods that everyone else was eating (especially the main dish which was always some kind of meat). My stepmother would respond, in a sort of disgusted tone of voice, "Just ignore her, she's weird." My father would remain silent - he did not intervene. Also, probably out of concern for my health, my stepmother's relatives would ask why I had such dark circles around my eyes (I always had huge dark circles around both eyes). My stepmother would respond in a snapping tone of voice, "She's always been that way." End of conversation. Again, my father would remain silent - he did not intervene.

As a young adult, I didn't "get it" that I really did have to eat differently than other people. My mother often told me that I had outgrown the MSUD (meaning that it was no longer necessary for me to keep a special diet). And I mostly believed her, although I did have some doubts. Consequently, I periodically banged into walls (figuratively speaking) throughout my twenties; I would get sick, but not realize that it had anything to do with my diet or the MSUD. At the prompting of my therapist, I decided to have a skin fibroblast study done, to see if I did still indeed have MSUD. I was 30 years old at the time. The re-diagnosis confirmed that I had MSUD and that my metabolic rate for the three offending amino acids was only 5% the rate of normal. At the age of about 35, I met with a dietitian (for the first time as an adult). She was very instrumental in helping me with my diet, and she also referred me to a doctor who is very knowledgeable about MSUD. I am forever grateful to her for this. She was also the person who mentioned that there was a support group for MSUD and that they had a newsletter.

Curiosity got the better of me, as it always does, and one day I searched the web and found the site for the MSUD Family Support Group. I printed out the subscription form and mailed it in that very same day. A few days later, Joyce Brubacher called me . . . and we talked and talked. I've very much enjoyed getting to know Joyce and I'm happy to have found this group.

In terms of diet, the foods that I can tolerate have changed over time. As a child, when I was very active and growing so much, one of my favorite things to drink was big glasses of chocolate milk (with tons of *Nestlé Quik* in it). I would NEVER do that now! I also used to drink milk shakes up until my late twenties, but I had to stop doing that when I got to be about 30 since it began to almost make me pass out. As a young adult, I could eat pizza,

but I am no longer able to do this since the cheese no longer breaks down in my mouth - it's just like trying to eat bubble gum.

Although my metabolic rate, for the three offending amino acids is only 5% the rate of normal, I have never had to use MSUD formula in my diet. I recently learned that MSUD formula did not even exist when I was a baby. In fact, MSUD formula was not commercially produced until I was a teenager. One of my safeguards for holding the MSUD in check is to always eat in small quantities - that way, if I do have a bad reaction to something, at least it won't be as bad as if I had eaten a larger portion. Also, there are foods I can tolerate (such as milk, eggs, and cheese) as long as I eat them in small quantities and in combination with other foods; but I do not tolerate them very well if I eat them alone or on an empty stomach.

My diet, which works very well for me, is the result of years and years of experiential learning, none of which I've done in an exacting, quantifiable manner. Rather, it's been a process of trial and error coupled with careful and intuitive observation.
I have worked with therapists for many years to heal from a myriad of emotional and psychological scars, most of which are related to living with a life threatening disorder and growing up in a very abusive family. I am currently in therapy to work on issues related to the MSUD.

Living with a life threatening illness isn't all bad - it has had some very positive impacts on my life. For one, I feel passionate about life and wake up excited to see a new day. Having come so close to death, hundreds of times over, has shifted my baseline of measurement as to what is difficult and what is easy in life. Things that other people get all worked up about tend to not phase me very much. Also, the extra challenges in my life have led me to develop a good sense of humor, as well as to be resourceful, self-nurturing, patient, persevering, imaginative and creative.

When I was 37, I became very ill and did not fully recover for about eight months. Exercise was absolutely essential to my recovery. I joined a fitness club and began a regular routine of aerobics classes. At first, I could just barely manage to do this without passing out during class. But gradually my health got better and better. Since then I continue to go to aerobics classes two to three times a week and eat very carefully. I think that exercise helps the body get rid of toxins more quickly than it otherwise would. For someone with MSUD, this is an important consideration.

When I had become so ill at 37, I was too weak to continue working full time, and I was granted a reduction in time to 90%. I was a supervisor at that time. In February of the following year, I received notification that I would soon be demoted - that I would no longer hold a supervisory position in the office. This was very upsetting to me and I decided to contact a lawyer. I found a lawyer who agreed to take my case and we proceeded to work together for about the next 4 months. With my lawyer's assistance, I filed a grievance against my supervisor, charging discrimination based (in part) on disability, and subsequently I won reinstatement of my supervisory duties. Before working with a lawyer, I had never thought of myself as "disabled." I had simply thought of myself as someone with MSUD. Through working with her, I learned about the laws that protect people with disabilities, such as the ADA, FMLA, and the Rehabilitation Act. Working with my lawyer was extremely transformative for me. It amazed me that someone would actually protect me due to the MSUD. I bonded quite strongly to her and completely inundated her with thank

you notes, flowers, and artwork (including a veritable flock of origami birds). Working with my lawyer has been one of the best experiences in my life, and it is one that I will always carry with me. In the words of Tracy Chapman, "I've met angels."

In closing, I'll give a brief overview of my educational background. I graduated from high school at the age of 16; I was an advanced learner and hence I was able to skip a full year of high school. I started college at the age of 18 and went on to earn a B.A. degree in studio arts with a minor in mathematics. After finishing my undergraduate degree, I entered graduate school in architecture and completed 3 years of full-time study in that field. I did not complete that degree primarily because of financial difficulties, and secondarily because I believe that truly fine architecture is a lost art. This September, I have begun graduate school in computer science and engineering at the University of Minnesota. I'll take one class per quarter and continue to work full time designing relational databases and web sites.

Art has always been very important to me. There are times when it serves as a container for emotions (such as panic and terror) that would otherwise completely overwhelm me. Art has literally been my lifeline at times when I was terrified I would die.

I welcome people to get in touch with me and I look forward to seeing all of you at the 1998 symposium. Be well.

SHARING - LYDIA WEBER, EDITOR'S NOTES

Details

Written by Joyce Brubacher

Published: 20 July 2009

The following letter was written by Lydia, daughter of George and Naomi Weber from Ontario, Canada. Since they are a Mennonite family, you may notice a quaint mixture of Pennsylvania Dutch and Canadian expressions in Lydia's letter. She candidly expresses her feelings about her disease and reveals some of the questions and issues faced by teenagers with a disorder like MSUD. The letter was written in early August 1997 when she was fourteen. She was the first to respond to my request for letters from teens and adults with MSUD. Lydia's mother wrote that Lydia left to work for an aunt after she had written this letter. Her formula was weighed out in advance for each day and she was responsible to mix and drink it.

Lydia's brother, Benjamin with MSUD, turned 17 in October. His mother wrote that he can handle hard physical labor even during warm weather. They drive horses and buggies and

Benjamin is not afraid to drive the young, not too well-broken, horses. He enjoys farm work, but reading and writing are hard for him. He is a little self-conscious about eating the low protein cake his mother sends along for him, whereas his younger brother Noah, with MSUD, loves desserts better than vegetables. Naomi wonders if eating so many calories and getting barely enough leucine could cause Noah to scream in his sleep.

The third boy with MSUD in the Weber family, Ishmael, died rather suddenly at the age of four years. He developed a fever and began vomiting after becoming overheated and exhausted on a hot day. This was in August of '82 and an account of his death was related in the Weber family history printed in our April '84 issue of the Newsletter. That family history also told of Benjamin's puzzling diagnosis. His father took Benjamin to Toronto to be tested on the day of his birth. His test showed negative, so he was taken home again. However, Ishmael became sick when Benjamin was born, and Naomi was very busy caring for the sick boy. It wasn't until Benjamin was eight days old, and would no longer suck, that they realized something was seriously wrong with Benjamin. A test for ketones showed very positive. He was taken to Toronto and diagnosed with MSUD. So this family has had many varied and trying experiences with MSUD. Thankfully all three MSUD are now doing quite well.

SHARING - LYDIA WEBER, HER LETTER

Details

Written by Lydia Weber

Published: 20 July 2009

Dear MSUD friends,

I will start with me. I am thirteen years old. I too am MSUD. I feel satisfied about it, because I learned to accept that I may not eat everything the others do. I have two brothers that are MSUD. I will name them later. I am glad to have two MSUD brothers although one died. I will name him later. I hope I do not get too sick, although sometimes I linger at that. I get severe stomachache and headache. Sometimes I believe Mother and Father don't like me. Mother and Father and sisters and brothers just don't know me. They don't understand, that's why it is so hard to get going with Susannah and Mother. The same with the boys. I never did dare to talk to Father. The boys, they don't understand me, I am quite sure. I guess the reason is because I am scared to talk to them.

If you ever want to meet me, you will have to be prepared to see brown hair, blue eyes, glasses, quite high, and a little slim. I am a healthy MSUD girl and I have good teeth. I think I grew, because when I was six or seven, I cut an apron belt because it was too long - and now it is too short. Mother and I once talked of MSUD. Mother said it was not the parents' plan that I must be different. It was all in the Great and Almighty Power's hand.

We have ten *(living children)* in the family.			
	John	20	Jan. 25, 1977
MSUD	**Ishmael**		**Born May 20, 1978**
			Died Aug. 17, 1982
	Eli	18	Apr. 21, 1979
MSUD	**Benjamin**	**16**	**Oct. 5, 1980**
	Susannah	15	Dec. 14, 1981
MSUD	**Lydia**	**14**	**Aug. 11, 1983**
	Samuel	12	Aug. 11, 1985
	Edwin	9	Feb. 21, 1988
	Sarah	7	Oct. 26, 1989
	Lucinda	5	Jan. 22, 1992
MSUD	**Noah**	**3**	**Nov. 16, 1993**

I like when it rains - the things can grow. The things I like to eat are low protein cake, potatoes, fruit, a little peas, corn, tomatoes, strawberries, raspberries, carrots, rhubarb and grapes. The things I don't like to eat are plums, flour, protein, cake, cheese, sausage, meat - because those I may not eat at all. Then there is milk, I may not drink. I may have water, tea, and all sorts of drink!

I like to watch things grow on a farm. As you know, we live on a fairly big farm. We have many trees, a short lane, a white house with a green roof, a brown-gray-red barn and a gray wood shop with a green door where they make rocking chairs. Once an old couple lived in the doddy house *(separate quarters for an older generation - usually grandparents)*, Samuel and Hannah Horst, but they died when they were very old. Now I sleep in the doddy house and my three sisters do, too.

We sold many strawberries. We also have four gardens and three rows of strawberries in the first garden and two rows in the second garden. In the third, we have flowers, peas, strawberries, onions, cabbage, red beets.

I like formula. I put Ketonex 2 (275 gm) and Polycose (150 gm) in and then orange drink, then add water *(this is divided with Ben)*. I also bake for the MSUD children on this farm in our household.

I like the evenings with the family at hand. I am tired. Today we did a lot of Monday work since yesterday was Sunday. Tonight we want to go to the field. The sun is shining. I am at the table, rewriting this to you. Yes, true enough. How I wish you would write to me. Tomorrow is Tuesday, August 5, 1997. I will go to Uncle Joseph Hoover's to help them. Eva Dyck is with us. I enjoy it and hope she enjoys it. The boys are out toiling till the day is done. This reminds me of a song: "The time for toil is past and night has come..." "Life is like a mountain railroad..." is another song.

I like to have brothers. When Noah was born, I felt very pleased and wished with all my heart he would have MSUD! How I hated when Mother and Father said they don't want him to have MSUD. Then later when Noah got sick, I realized I shouldn't have wished something like that! Now that Noah is three, he is a busy little brother. To us, he is an enjoyable boy. Now years later, I am thankful to have Noah as MSUD, but even if he would not, I would still enjoy him. You don't know what it means to me to have Noah as a partner in MSUD. Now, you know, Noah is three and still drinks from a bottle. What can we do?

When I crack eggs to bake a cake for the healthy ones, I think, "Now, why must I? I won't even have a husband if time goes on. I might be an old maid. Who knows? I may not even eat the dough - why then must I bake for them?" I don't always appreciate to bake, but if it is my duty, I might as well accept it.

How do you feel? Do you sometimes feel sick? I would like to write to somebody my age who has MSUD. I would like to write to one of you. I would like if you would tell me what you like. You wouldn't have to put it in the Newsletter, write just like a pen pal.

If someone would like to be a pen pal to Lydia, e-mail msud-support@characterlink.net

SHARING - SAMANTHA'S SYMPOSIUM '96 SPEECH

Details

Written by Samantha Whitney

Published: 20 July 2009

In the last issue of the Newsletter, the Spring/Summer issue, I mentioned I would print Samantha Whitney's talk in this issue. Since she shared so candidly with us as

a young adult with MSUD, her speech fit quite well in this Sharing Section. She spoke following her mother's speech at Symposium '96 during the presentation of "International Aspects of MSUD."

My name is Samantha Whitney and I'm from a town called Nowra in Australia. I would like to thank you for inviting me here to America. I've always wanted to meet people with the same illness or condition as me and now I have that chance - so thank you.

I'm twenty years old and I've had maple syrup since I was one week old. I'll talk about how I feel about the condition. Sometimes it really gets me down because I have to drink this horrible formula. I'm wondering if any of you with MSUD have the same feeling about your formula. I will have to get around to know some of you, and maybe I could try some of your formula. Mine comes from England and is called Maximaid.

It also gets me down how you have to stick to a very strict low protein diet as most of you with MSUD can relate to. I don't know about you, but when I'm feeling sick, the last thing I want is my formula. It makes me feel even worse. I have had to sometimes boost it up a little, and it just tastes horrible!

Would most of you with MSUD agree that the low protein diet is plain and boring and . . . yuck? That is what I think. Sometimes I would just like to have a meat pie or a hamburger. But I know I can't, as it would probably kill me. I mean you probably feel like that every now and again. I don't know if any of you with MSUD feel the same when you're asked why you can't eat meat. I know I hate it when you have to go and take your formula and explain why you can't eat meat when you're staying at someone's house, like a friend's house.

I also dislike the fact that there is no one else to talk to about how I feel with this condition and there is no support in Australia for MSUD. It is like no one knows what you are going through or what it is like to have this condition.

The other thing I hate or dislike - and I'm sure some of you with MSUD can relate to this feeling - is feeling nauseated and not being able to be sick or being sick and not being able to stop being sick. I really hate that. I haven't had as many hospital stays as I did when I was younger, however, I still get hospitalized every now and again.

I know I haven't said anything positive about the condition, as there really isn't anything positive about it. The only positive thing is getting the chance to come over here to America and meet other maple syrup people the same as me, which I really love. I mean it is great to get around and talk to people who have got the same thing as I do. It is just really good.

I'm counting on doing a two year course at TAFE called Youth Diploma. It is a full course and I love it. It is something I really want to do. I believe here in America you call it college, whereas we call it TAFE in Australia. I have a great set of friends at TAFE who accept me for who I am, and it is really good. They don't look at me as being different, so I'm really happy about that.

And I guess when I look into the future I see myself married and hopefully with kids and happy. However, there is always hope in the back of my mind that there will be a cure one

day for all the maple syrup people. I would like to thank you for your time and thank you again for inviting us to America.

SHARING - A WISH GRANTED

Details

Written by Sandy Bulcher

Published: 20 July 2009

Shortly after the 1996 MSUD Symposium in Columbus, Ohio, I received a phone call from the Columbus branch office of a national wish foundation, *A Special Wish*. Apparently, one of their volunteers had seen the *Columbus Dispatch* article about our son Jordan that mentioned MSUD and the Symposium. The caller proceeded to say that because of the information in the article, Jordan may qualify to have a wish granted. Immediately I got a sick feeling in my stomach. My perception was that wishes were granted only for terminally ill children.

My initial response was to defend MSUD. For the most part, I explained, MSUD children were doing better than ever. She explained that to have a wish granted the managing physician needed to sign a form stating that the disease is potentially life threatening. I was surprised to learn this, but still felt uneasy about the whole idea. I realized that I viewed Jordan more as a happy boy with a special diet than as an ill child, so it was difficult to believe that he could qualify. However, within a few months, the paperwork was complete and off we were to Orlando, Florida.

A Special Wish provided a long white limousine to take us to the airport. Tyler (our 10-year-old) and Jordan (age 7) had never been in a limousine before, so this was really fun for them. We arrived in Orlando at 1:20 p.m. and were greeted by a volunteer holding a sign with Jordan's name on it. After getting our rental car, we drove twenty minutes to *Give Kids the World* Village.

Give Kids the World is an independent, non-profit organization dedicated to providing six-day, all expenses paid family vacations in central Florida for children with life threatening illnesses whose wish it is to go to Disney World, MGM or Universal Studios. The organization is supported by donations from many corporations and hundreds of volunteers. Families are referred to *Give Kids the World* by more than 250 foundations, hospitals, and hospices worldwide.

The Village, located on 350 acres of land, was opened in 1989. We stayed in one of the beautiful two bedroom vacation villas. There was much to see and do at the Village. Meals were provided in the Gingerbread House. There was a wheelchair accessible, heated

swimming pool, playground, fishing lagoon, nature trail, a castle (where the boys played video games and never had to put a coin in), an ice cream parlor, and a carousel.

Friday morning, October 18, 1996, we started our first full day in Orlando. We ate breakfast in the Gingerbread House with the other families. It was difficult to see some of the children who were obviously very ill, and my heart ached for them and their families. There was a wide variety of food served cafeteria style, and we always found enough food appropriate for Jordan's diet. We spent most of the day at MGM Studios. In addition to payment of all travel expenses and admissions, we were given a predetermined amount of cash to spend on food outside the park and for souvenirs. The boys really enjoyed the Indiana Jones Stunt Show and the Honey I Shrunk the Kids Park. Jordan wore a *Give Kids the World* button which proved invaluable. We were able to enter the handicapped entrance and board rides or attractions before other park guests. Most park employees recognized the button and paid special attention to him.

Saturday we spent the entire day at the Magic Kingdom. The boys really enjoyed Thunder Mountain Railroad (roller coaster) and the Haunted Mansion. It was a crowded day, but because we didn't have to wait in lines, we were able to see and do many things. The driver of the Jungle Cruise boat ride spotted Jordan's button and asked him to steer the boat. He was so excited. In the evening, we were given front row seats for the Electric Light Parade. Sunday, we spent several hours at Epcot Center and then relaxed around the Village pool in the afternoon. We were still exhausted from the previous day's adventure at the Magic Kingdom. Sunday evening we went to Wild Bill's, where we ate dinner while we watched country dancing, Indian dancing and rope twirling. At 9:30 that evening, Mayor Clayton, the Village Mascot came to our villa to "tuck-in" Tyler and Jordan. Mayor Clayton was a very large bunny character who spent 15 minutes teasing and playing with the boys. They giggled so hard that it took them a long time to settle down and fall asleep.

Monday morning we ate breakfast in the Gingerbread House with the Disney characters and then went to Universal Studios. The highlight of the day was petting the St. Bernard dog that played Beethoven in the movies. While we were waiting in a long line for lunch, the attendant saw Jordan's button and we were escorted to the front of the line and seated immediately. Everyone at the Village and the Theme Parks were very generous and treated us with extra kindness. Monday afternoon we went to Gatorland. We expected to see a few alligators, but instead there were alligators everywhere! I think Dave and the boys enjoyed them more than I did. That evening we went to a pool party at the Village with the Shamu character from Sea World.

Tuesday morning we packed up and headed back to the airport. We were greeted at the Columbus airport by a limousine driver wearing a tuxedo and holding a sign that said JORDAN BULCHER. Jordan felt so special. Every detail was thought through and well planned by both the Kids Village and the Wish Foundation. Undoubtedly, it will be our most memorable vacation. We felt very fortunate to have experienced such a great trip and are thankful to all who made it all possible.

The national headquarters of *A Special Wish Foundation* is also located in Columbus, Ohio. To have a wish granted, a child's physician needs to determine that the disease is potentially life-threatening, the child must be under 20, and live in the United States. Only

one wish per child is granted. Anyone can call the national office of *A Special Wish* at 1-800-486-9474 and they'll determine which office in your area will grant the wish.

SHARING - OUR TRIP WITH A SPECIAL WISH

Details

Written by Renee Eck

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After an article was published in the Columbus newspaper about the MSUD Symposium, Sandy Bulcher and I were contacted by the organization, *A Special Wish*. This organization grants wishes to children who are terminally ill, or who have a life-threatening condition.

Cory wanted to go to Walt Disney World in Florida for his wish. All four of us got an all-expense paid trip to Disney World. This included air fair, accommodations, food, tickets to every park, coupons for free meals, dinner at a dinner theater, and a night out for the parents. There was a lot more of this type of thing, but you have to have experienced it to get the full effect.

We took our trip in February '97. This is a colder time of year in Ohio, so we thought it would be a good time to get away. We left the Columbus airport and landed in Orlando, Florida about two hours later. A gentleman was there to meet us. He and his wife helped us with our luggage and took us to pick up our rental car. It was quite a big airport and we would have been lost if they wouldn't have been there to help us. The gentleman also gave us directions on how to get to *Give Kids the World*. That is the place where just about everyone stays when an organization sends you on a trip like this.

At *Give Kids the World*, that is just what they do. You can get anything you may need from the people who work there. I received a wheelchair for Cory to use while we were there at the Village. I didn't need it at the parks, because every park made sure we got one. When I had to make Cory's formula, I just went to the front desk and asked to use the blender. I would use it right away and take it right back for someone else to use. If you wanted to have some of your pictures developed while you were there, they would send them out for you. (You did have to pay for them.)

Give Kids the World is a very different place to stay. There's a Gingerbread house were you go every day to eat breakfast and dinner. You need to buy your own lunch. We ate breakfast at the Village, and a lot of times didn't need to eat lunch. We always picked up something for Cory. We had so much to eat, it kind of made you sick of food. There is a castle where you can play video games, play with toys, and also get your picture taken with

Mickey and Minnie Mouse. That was something the staff had you do to help you remember your great trip.

We went to every park - MGM, Universal Studios, Epcot Center and the Magic Kingdom. We had all kinds of coupons given to us at the Village. We got a free lunch or dinner for four at the Hard Rock Café at Universal Studios. Each and every park was great. We tried to see and do all we could while we were there. Of course, there wasn't enough time. We did get very tired, but we were only going to do this one time, so we just kept going.

We also got a night out at a dinner theater. There were three or four to choose from. We went to Wild Bill's. I thought that would keep Cory's interest longer. It was really fun. They danced and sang, had cowboys doing rope tricks, Indians dancing and doing all kinds of different things. It was really neat how they would sing for each course of the meal when they brought it out. Cory really enjoyed it. We did have a small problem here with the food. They really didn't have anything Cory could eat. I would suggest having your MSUD child eat before going to Wild Bill's and take some things with you for them to eat there. This is the only place where we ran into trouble with his diet.

On our last day, we went to the Magic Kingdom. On our way out of the park, we decided we wanted to eat out. I had been wanting to go to a Planet Hollywood Restaurant. There was one there and I just had to go to it. We stopped and walked up with Cory on crutches. (We left the wheelchair at the park.) I was asking a man who worked there if there was a way to get in, other than up the stairs. Just then another man came up to us and had us follow him. He took us up in the elevator and got us a table ahead of everyone else.

As the waitress walked away from our table after we had ordered, I asked Jim how much something was that he ordered. We did get a little concerned about the cost, after we saw the prices. We had no coupons for this place and had not been told that they would do anything for *Give Kids the World* kids. The waitress came back to our table and asked us to please reorder. She said that "our dinners were on Planet Hollywood" that night and that included drinks and dessert. Also they brought us three plates of different appetizers to try. Their servings are very large, so we couldn't eat all of our dinners. They kept wanting us to take dessert with us. We were so full, dessert didn't even sound good. They gave us two tee shirts and gave Cory a stuffed gorilla wearing a Planet Hollywood tee shirt.

We had such a great time; I hope everyone who has a child with MSUD will be able to go. With all the many bad times we have had with Cory and this disease, this was one of the "up times" we will never forget. I can't thank *Give Kids the World* and *A Special Wish* enough for this very good time our family spent together with so much never-ending fun.