# VOLUME 14-2

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# SYMPOSIUM '96 REVIEW

**Details**

Written by Joyce Brubacher

Published: 20 July 2009

The MSUD Symposium, held this year on June 20 through 22 in Columbus, OH, is history. History should be recorded. So this issue of the Newsletter will attempt to help those who attended the conference to recall some of the information and good times, and also share it with those who could not be there.

Dave and Sandy Bulcher did a super job, planning and putting on such a great, well-organized event. They are still involved in follow-up projects and tying up loose ends, such as passing on information and material to the PA group for the '98 Symposium.

Tish Fuller took notes on the talks given at the Symposium. I was able to listen to the taped speeches and add some information to her notes. Most speakers used visuals and handouts, which complemented the presentations. In the following summaries, I attempt to cover highlights of the speeches.

Due to the good response from the families during and since the Symposium, I have more material than will conveniently fit into this Newsletter. So the following summaries only cover the speeches given the first full day (Friday) of the Symposium. I expect to cover Saturday's speeches and activities in our next (spring/summer) issue. That issue will include material on sibling issues, nutrition, self-esteem and descriptions of MSUD treatment in Chile and Australia.

# MSUD: FROM PROTEIN TO GENES TO THERAPY, THANKS TO THE FAMILIES!

**Details**

Written by Joyce Brubacher

Published: 20 July 2009

**Presenter: Dean Danner, PhD**

**Since 1973, Dr. Danner has been studying mutations that cause MSUD. He is a professor in the Dept. of Genetics and Molecular Medicine at Emory University, Atlanta, Georgia. He sincerely thanked the families for their help and cooperation which makes gene therapy possible. He answered technical questions in a relaxed and easy to understand manner.**

### 1) What do we know about MSUD?

The function within the cell that is not working is the enzyme, branched-chain alpha-keto acid dehydrogenase (BCKD). It breaks down the amino acids in proteins. When it doesn't work, the branched-chain amino acids (BCAA) and their keto acids (BCKA) build to toxic levels in tissues and fluids.

There is no racial or ethnic preference. Dr. Danner has cell lines from China, Japan, Africa, Saudi Arabia and Turkey.

How often does it occur? The incidence is one in 176 in the Mennonite community, 1 in 100,000 in Georgia and 1 in 185,000 in the general population. There is a common mutation in the Mennonite community, but no other common mutations have been found. There was a different mutation in each of the other families he tested.

### 2) What have we learned in molecular genetics?

BCKD is found in the mitochondria. Every cell of the body has mitochondria except red blood cells. The amino acids have to get into the mitochondria and the BCKD must be available to break down the amino acids to produce energy. The chemistry is very complicated. The enzyme is in active form at only 40% in the kidney cells, 3% in muscle cells and maybe 30% in the brain cells.

### 3) How do we detect mutations within families?

Dr. Elsas used the breath test at the Symposium in Toronto. It tests the ability to oxidize leucine. Many of those analyses are still being done because it takes a long time.

From blood samples, Dr. Danner transforms white blood cells so they grow in culture. It takes 6 to 12 weeks until they are ready to study. After that he does an enzyme activity and a Western Blot to look for the presence of the $E_2$, $E_1$-alpha or $E_1$-beta protein to see which is missing. (The mutation in MSUD can occur at any of these three subunits of the enzyme complex.) He also looks at the RNA which is produced from those genes and then checks the parent cells to see if they really got this change from Mom and Dad. Using illustrations Dr. Danner briefly explained the complicated tests he uses to find the defective gene in individuals.

The combined information from several laboratories has identified 20 mutations on the $E_1$-alpha subunit, 7 on the $E_1$-beta subunit, and 11 on the $E_2$ subunit. One parent may have a different mutation from the other but in the same subunit.

**4) What has this knowledge done for MSUD families?**
Since MSUD is now treated with the protein restricted diet, the old clinical classifications are useless. We need to determine the best treatment for a specific type of mutation.

Prenatal monitoring is possible when we know the mutation within that family. Any other pregnancy in that family can be monitored by enzyme assay or DNA analysis. Also other relatives can be tested for the mutation but only at the DNA level.

Thiamine responsive MSUD is difficult to identify and is not well understood. It is not a deficiency of vitamin $B_1$ but requires pharmaceutical doses of 100 to 1000 times the normal amount of thiamine ($B_1$). Some clinics give it to all the MSUD patients because of the possibility it might help and it is not dangerous. Patients proven to be thiamine responsive have an $E_2$ mutation.

Two other things we've learned are that newborn screening is very important in averting the consequences of MSUD, and that the diet is necessary for life.

**5) What questions remain? Are there new questions?**
We want to identify all the mutations. The technology is there but we need hands and dollars to do it. As a geneticist, I want to know why these changes cause a dysfunction so better management can be provided. We want to relate genotype to therapy and also identify which is the most important tissue for BCKD function in humans, so we know which tissue to target for gene replacement therapy.

The new problem is maternal MSUD. Being on diet, young women with MSUD are capable of reproduction. How do they nurture a fetus who needs a high protein diet to develop? It is necessary to identify when, in the embryonic development, the BCKD is turned on in the fetus. There is a chance the fetus can help the MSUD mom metabolize the BCAAs during pregnancy so she does not get sick. Only one pregnancy of a woman with MSUD has been reported in medical literature and she had a normal baby.

**6) Future Goals:**
Enzyme replacement is probably not possible, but gene replacement or organ transplant (like kidney) is a possibility. However, after identifying the best tissue, how do we deliver the corrected gene and how often will replacement be necessary? There is hope that gene replacement could be possible - it is possible now in the lab - but it won't happen tomorrow. Dr. Danner is developing an MSUD mouse. An animal model is needed to study the neurological complications in gene replacement therapy and to detect during pregnancy when the fetus can begin to handle the leucine overload so the MSUD mother can increase dietary protein.

**Questions answered:**
Enzyme activity is no gauge of phenotype or how well protein is tolerated. One carrier father has 2% activity level and is perfectly normal. Both carrier parents of two children severely affected have 100% enzyme activity. A child with severe clinical signs of MSUD can have activity level at 20 to 30%. Another child can be doing very well with less than 1% enzyme activity. We don't know that much about the enzyme activity. Other genes interact and persons respond differently.

Are persons with MSUD missing something by not getting energy from protein? Biochemically it takes more effort to make energy from protein than from carbohydrates. So most energy comes from carbohydrates, except when they are lacking, then it comes from protein. Muscles may fatigue more quickly because they cannot activate the enzyme to get rid of toxic products. Muscles have only 1 to 2% of the active form of the enzyme.
In comparison to PKU research, we are not as advanced. PKU involves one organ, the liver, and 1 gene. MSUD involves 3 genes, and we still do not know the organ to target.

Questions were asked of Dr. Danner about how his research was funded. All of his research funds now come from NIH and it is getting tougher to get funding. Companies will not fund research for such a small group of patients as MSUD. We can write to our Senators and Congressmen to continue and increase the funding for NIH. It is urgent that funding continue to ensure further research efforts. Dr. Danner would gladly provide anything needed to raise funds for his research.

# QUESTION &AMP; ANSWER SESSION

**Details**

Written by Joyce Brubacher

Published: 20 July 2009

**Panel members: Richard Allen, M.D., Anna Marie Schaefer, R.D., MPH., Dean Danner, PhD.**

The issue of classifications was discussed at length. The original classifications included classic, intermediate, intermittent, and thiamine responsive. These descriptions are based on the clinical differences, leucine tolerance and response to thiamine. They do not necessarily correlate with the enzyme assays. The consensus seems to be that with so many unknowns and variations metabolic classifications are inadequate. Determining the mutation may be a better way to indicate type of MSUD. Hopefully a mouse model will help in determining differences.

One mother asked about how to increase the calories in the diet of her 14 year old boy who is small and thin for his age. If they add high calorie supplements, his appetite is affected and he won't eat. Children with MSUD vary in size and weight and it is hard to know what is genetic and what may be the effects of nutrition.

A mother said her daughter was turned down when she wanted to give blood. Both Dr. Allen and Dr. Danner said there was no biological reason a person with MSUD can not be a blood donor. However, a knowledgeable person needs to be contacted and not just the person drawing the blood.

Doctors in Australia told a mother TPN is too expensive and lipids do the same thing. Dr. Allen explained that TPN has been used all over the world for many years, but TPN for

MSUD is revised with the branched-chain amino acids removed. Massive doses of lipids can cause other metabolic problems. Individuals with MSUD need the protein in the TPN.

The best levels to maintain in a child with MSUD depends on whether you are trying to attain a preconceived number or are based on how the child reacts. Would the child grow better with a little higher level? The labels are wrong. Whether the child is classic, mild or intermittent is not important and can lead to confusion. A child that is not gaining weight or losing weigh is catabolic regardless of how "good" the blood level is. Ultimately the child needs to be kept at the level that will keep that child healthy and growing.

There are various effective treatments which vary with the individuals and different clinics. But when a child starts showing neurological (brain stem) involvement - not cerebral edema - Dr. Allen gives that child TPN. This usually results in a rapid and full recovery without any other treatment.

A mother stated there was no doctor or center involved in treating MSUD near them. Dr. Allen says we need to find a way to help physicians and pediatricians understand that MSUD is a real disorder, and there are newer and different ways to treat these children. However, it is still remarkably better than it was 10 years ago.

# ADHD IN SPECIAL NEEDS CHILDREN

**Details**

Written by Joyce Brubacher

Published: 20 July 2009

**Presenter: Chip Kobe, PhD.**
Searching data bases, Dr. Kobe found only 2 articles on the intellectual and behavioral functioning of children with MSUD. He addresses Attention Deficit Hyperactivity Disorder (ADHD) recognizing that it is secondary to the medical issue in MSUD.
The time of diagnosis and the kind of metabolic control play a part in the developmental and behavioral aspects of MSUD. When in good metabolic control, behavioral issues are probably based upon that child's personality, but when out of control the behavioral issues may be a part of metabolic control and not normal functioning. Compliance with the special diet is also involved in MSUD.

Some areas in which psychologists can be of help:

- Developing behavioral modification approaches
- Improving adaptive skills
- Addressing parenting stress
- Developing child's self confidence and self esteem

A neuro-development disorder is characterized by:

- Deficits in sustained attention for mental tasks (academic vs. Nintendo)
- Difficulty inhibiting behavior - impulsive, not thinking before they act
- Problems regulating activity level - hyperactivity
- Impairment in the ability to use rules and instructions to guide their behavior
- Decreased ability to work toward long-term goals

Inattention, impulsivity, and hyperactivity, the core symptoms in ADHD, necessitate changes in lifestyle - parenting and educational approaches and sometimes medication. These children are prone to relapse. Behavior management takes more time, effort, patience and persistence than with other children.

Three to five percent of school age children are diagnosed with ADHD. Currently there is a 3:1 ratio of males to females but 6:1 males are being treated. Many females are being missed. ADHD is found in all countries and ethnic groups.

There are few differences in core symptoms in the genders. Females have fewer conduct problems or aggressive behaviors, more learning problems, trouble focusing their attention and more symptoms related to anxiety and depression.

There are biological, social, psychological, and developmental factors which influence how children develop and behave. There are also issues involved related to their cognitive development, memory processes, their personality, and also perceptual and motor skills.

**ADHD primary symptoms:**

1. Inattention- impairment that is not typical for their age, problems with attention to detail, don't listen when spoken to, difficulty organizing tasks, easily distracted, forgetful
2. Hyperactivity- fidgety, trouble remaining in their seat, can't play quietly, seem to be on the go, excessive talking
3. Impulsivity- blurt out answers before questions are completed, difficulty waiting their turn, interrupt others

These affect a child's social skills. Other children pick up these symptoms first, and begin to exclude ADHD children from the group. These children are perhaps not as predictable and don't follow the same routine as the other children. We need to enhance their social skills.

**May have co-existing problems:**

- Psychiatric disorders
- Opposition or defiant behavior
- Conduct problems
- Depression - anxiety
- Having a lot of physical complaints with no medical cause
- Learning problems in school (MSUD are at risk)
- Excessive variability in performance
- Not as productive as expected to be
- Lower than expected levels of achievement
- Emotional problems
- Issues of self-esteem

- Low tolerance for frustration - over-react to situations
- Peer rejection
- Little regard for social consequences
- Immature play and social interests
- Don't read their social environment - misinterpret actions of others

Parents can do a functional assessment of their child. Be sure to consider the events or situations that occur prior to the behavior. It may be simply a way to get attention or there may be another function to that behavior. Don't jump to the conclusion it is related to ADHD. When does it happen? Is it an issue of metabolic control or are there other stressors?

**Behavior Management of children with ADHD:**

- Give immediate feedback and consequences for their behavior.
- Give more frequent and higher level feedback and consequences in managing their behavior.
- Institute behavioral approaches that use more powerful consequences (star charts, etc.).
- Encourage and praise more often.
- Always use positives before negatives.
- Be consistent over time and in different settings.
- Anticipate problems to limit the tendency to over-react to the child's problem behavior.
- Keep disability perspective in relationship to the child's behavior (avoid anger/embarrassment).
- PRACTICE FORGIVENESS! (for child, yourself, and those who misunderstand the situation)

There are effective and scientifically proven treatments for ADHD. Medicine is now accepted along with behavior modification, but there are no studies that look at the use of medications, such as Ritalin, to treat ADHD symptoms in children with MSUD. The traditional medications used for ADHD do have the side effect of appetite suppression. A new medication may have fewer side effects but is not widely available at this time. Using antidepressants may be an alternative.

There is a support group called Children and Adults Having Attention Deficient Disorder (CHADD). Most communities have a CHADD chapter which is a good resource.
Parents need training in child management approaches. Some of the intuitive things parents do don't work with ADHD children. Teachers need training in classroom management. They need to know which children have learning disabilities and which ones just need a little extra help.

Parents may need to be an advocate for their child to access proper services in schools. Special education is important for some; a multi-faceted evaluation is important to determine qualification for special services. There may be psychologists or other services within your medical center which can help with these issues.

**Unproven/Disproven Treatments:**

- Dietary management for ADHD
- Megavitamin/Orthomolecular Therapies

- Sensory-Integration therapy for ADHD
- Chiropractic manipulation
- Ocular motor exercises/optometrics
- Traditional play therapy
- Neuro-feedback (EEG biofeedback)
- Self-control training in clinics
- Social skills training in clinics

There are no easy answers. But if you can document the effect upon the child's functioning, you can make substantial changes in the child's overall behavioral functioning and learning. These core symptoms do follow a good percentage of persons into adulthood. However, adults seem to learn coping strategies and find jobs, careers, and educational interests where symptoms have a lesser effect on their daily functioning. Some adults still benefit from treatment.

# STATISTICS OF THE 1996 SYMPOSIUM

**Details**

Written by Sandy Bulcher

Published: 20 July 2009

It's been a few months since the Symposium and our life is getting "back to normal." Thanks to all of you who wrote and shared your thoughts. It was wonderful to see so many old friends and meet so many new families. I wanted to share some statistics that I think you may find interesting.

1. 55 MSUD families from 4 different countries attended - U.S., Canada, Chile, Australia
2. 24 states were represented.
3. Pennsylvania had the largest number of families with 14.
4. 55 children and young adults with MSUD attended.
5. The oldest MSUD female was Cindy Blau, age 37, from Columbus, Ohio.
6. The oldest MSUD male was Darrell Deel, age 32, from Birchleaf, Virginia
7. The youngest MSUD child was Gina Basile, age 20 months, from Olney, Maryland.
8. 7 families had more than 1 MSUD child with them.
9. 33 siblings of various ages were present.

And finally, there were a number of professionals present from several different states. I was really pleased with the great turn out. Let's make the 1998 Symposium in Pennsylvania even bigger and better. See you there!

# APPRECIATION FOR SYMPOSIUM

**Details**

Written by Joyce Brubacher

Published: 20 July 2009

**Following are comments from letters received by Sandy Bulcher since the Symposium.**
*Young adult with MSUD (PA)*: I wish there would be an MSUD meeting every day.

*MSUD Mom (PA)*: It gave us fresh courage to come home and keep trying to fulfill our duties in caring for our MSUD child and the rest of the family.

*MSUD Mom (CO)*: It is great to be with people who walk the same path. At least we know we are not alone.

*MSUD Mom (Canada)*: The ambiance that was created by the presence of so many families (old and new) was impressive and most of all heartwarming.

*MSUD Mom (Canada)*: The input at the parents meeting helped shape the program and made the atmosphere congenial and warm. We loved it.

*Rosemary from Australia*: Sam & myself have been on the local ABC radio here talking about MSUD and the Symposium in Ohio. After traveling all the way to America and talking in front of the group, I must admit we sounded pretty professional, even if I say so myself!"

# A GREAT TIME

**Details**

Written by Joyce Brubacher

Published: 20 July 2009

**Sandy was too modest to pass on all the comments of praise she and Dave received. We have heard many. Anyone involved in putting on a meeting like this knows the huge amount of time, effort, and the mental and emotional challenge involved. Wayne**

**and I extend our admiration and deepest thanks to Dave and Sandy for an excellent job and attitude. Following are some of the comments we heard or received in letters.**

*Comments from Jeannie Gauvin*: This is the first time my teenage sons, Allen and Jason, and my husband attended the Symposium with me. We all agree it was the best vacation we ever had. The boys say they had an awesome time, liked the food and pool, and made friends with many of the other kids and adults. Allen loved helping sing with the group at the end, and Jason regrets not helping. It was great sharing this time with my family and being part of the larger MSUD family. There was a peculiar bond of understanding in the emotional highs and lows as we shared our common blessings and burdens. We all felt cared for, and cared about, accepted and loved! Mr. & Mrs. Bulcher did a marvelous job organizing this Symposium and especially addressing the issues with the older kids. We don't want to miss another Symposium if possible.

*Parent of a Teenager*: The long weekend charged our emotions, challenged our minds and certainly inspired us to move forward on some important issues and future planning. When I look at our grown up youngsters each doing their own special thing and reflect on years gone by, I simply start to cry - the mixed emotions of pride and sadness are just too much. They are truly "beautiful children." I think Sandy and Dave did a remarkable job in putting together such an impressive meeting for us and a great program for the kids. They were efficient, warm and caring. I do hope others appreciate their efforts and hard work as much as we did.

*Unsigned*: I think this was the best MSUD Symposium we had - such a large group. Was very interesting meeting these older MSUD people, to hear of their experiences. Also enjoyed meeting the people from Chile and Australia and to realize how fortunate we are for the treatment we have. These MSUD children taking part means so much; also hearing parents tell their experience means so much.

*Mabel Burkholder*: Definitely write up about Symposium in Newsletter - especially for the people who could not come. I really appreciated it when I was home with a sick child. I feel so good about the whole thing - meeting and talking with parents and getting new ideas from doctors. Thank you.

*Other comments*: "We came home with a new commitment and purpose." "The second day was so worthwhile. I really enjoyed Phyllis Acosta, the Sibling Issues, and the parent talks. Most of the speeches were so positive." "I was really touched by Herb Foster's sharing about his son's death."

**Renee Eck helped enthusiastically with fund raising and with other responsibilities at the Symposium. Below she shares some thoughts on the conference.**

I really like the booklet we all received in our folders that Dr. Allen had made up about everything on MSUD. I took it with me to the hospital on our next visit and it helped some of the other doctors that were helping with my child. I suggest taking it with you on hospital stays to help others who do not understand what all the problems are with the disease. And also if you have to go to a different hospital or see a different doctor. It was a real help to us.

I really enjoyed Dean Danner who talked on genetics. He was very open and very funny at times. Even though everything he talked about was not always easy to follow or understand. I think I got a lot out of what he was talking about. I THINK?

Even though I knew a little about the skit, "The Things People Say", I thought it was really good and should be printed in the Newsletter. The actors, Kathy Bachmann, and Dave and Sandy Bulcher couldn't have done a better job. They were GREAT!

Others speakers I really enjoyed were: Vicki Delaski on Sibling Issues, Emilio Amigo on Self Esteem, and the parent speakers, Loreto Ilabaca from Chile and Rosemary Whitney from Australian and her daughter Samantha.

After hearing the three international speakers, I don't think you could help but think about how much harder it is for them where they live. We do have an easier time with this than they do. I will think about them a lot, and I hope they keep in touch with the group. I'm sure that many of our thoughts are with them.

**Cathy Codner sent me a letter soon after the Symposium which included the following comments. Her daughter, Lauren is 10.**
The biggest effect the conference had on our family was on Lauren . . . and for that I will be forever grateful. For her, having MSUD and being different was always a negative feeling and her self-esteem always had a large chip in it. When we left the conference, we had a long talk. I'll summarize her thoughts in this way:

1. She enjoyed the conference.
2. She was a bit scared by what she saw and the effects that MSUD has on many of the children.
3. From that Lauren realized that her diet is extremely important, and she needs to pay more attention to it.
4. She became aware for the first time, by observing others, how behavior can change when someone's blood levels fluctuate. She could never understand before how I could tell that she needed to get something to drink, or she needed to rest, or whatever I observe. As the conference went on, and she observed others, things seemed to make sense to her.
5. Most importantly, Lauren felt for the first time that having MSUD made her special - and that being special is a good thing, not bad.

Lauren's attitude had become so much more positive. I can't tell anyone how important the conference was for us! Never having attended before I will not miss another one.

# HOSTING THE '96 SYMPOSIUM

**Details**

Written by Sandy Bulcher

Published: 20 July 2009

I had requests to write about what is involved in hosting a Symposium. I had mixed feelings about how to write the article. I didn't want the article to sound so overwhelming that no one ever volunteers again, but I did want to be honest and convey that it involves A LOT OF WORK - too many details to list completely.

Even though I never hosted a Symposium before, I was smart enough, though, to utilize valuable resources (Susan Witter, MSW, host of the '94 Symposium and Wayne and Joyce Brubacher). My husband Dave and I volunteered in June '94 at the Missouri Symposium. In August of '94 I called many area hotels and requested information packets. I narrowed the list of possible hotel sites using several criteria - location, size and number of meeting rooms, reasonable and adequate accommodations, etc.

In September, '94 I visited several hotels that met the criteria and finally decided on the Comfort Inn. After confirming Dr. Allen's schedule, I selected a date for the Symposium. January of '95 was the start of fund raising. I had never done any fund raising, so I first read several books from the library. I developed a fund raising packet with sample letters, receipt forms, and thank-you's and sent them to my fund raising assistants, Renee Eck and Sandy Kiel. From January to December '95, I sent out nearly 200 letters to formula companies, low protein food companies, local businesses, organizations, physicians, family, friends, and neighbors.

Renee and Sandy also sent out many letters requesting financial assistance. By December of '95, we had raised $13,000. Incidentally, several large organizations decided not to fund the Symposium, because their money "wouldn't benefit enough people." I also applied for a substantial grant from a local organization, which proved unsuccessful.

With most of the fund raising finished, my next hurdle was to finalize the topics and speakers. I reviewed many old Newsletters and published articles to decide upon meaningful topics and knowledgeable speakers. I also spent many hours on the phone getting referrals for local speakers.

The next few months were spent working on details, i.e. children's entertainment, child care providers, and mailings to the speakers and families. April '96 was a busy month as the Symposium was only 2 months away. More specifics about the Symposium were sent to families and speakers.

May and June proved to be the most hectic months of all, as there was many specifics, i.e. child care coverage, volunteers to make low protein snacks, accommodating needs of speakers, just to name a few. It was challenging to keep abreast of the ever-changing list of families and professionals attending. And finally, the day before the Symposium, I provided the hotel manager with numbers for meals and conference room seating. The evening before the Symposium we hosted Rosemary and Samantha Whitney (Australia) and Loreto Ilabaca (Chile) at our home. We really enjoyed this special time.

In spite of the large amount of work involved in hosting a Symposium, the experience was very rewarding personally. I learned a great deal and, hopefully, have grown through the experience. I'd be negligent if I didn't mention how helpful Joyce Brubacher was. I called her many, many times for ideas and suggestions. My husband Dave's organizational skills and knowledge of computer software were very helpful, also. And finally, a special thanks to our sons, Tyler and Jordan, for their patience during my many phone conversations.

# SHARING - LETTER FROM JENNIFER SAUNDERS

**Details**

Written by Julia Saunders

Published: 20 July 2009

June 22, 1996

Dear Friends of the MSUD Family,

Hello! This letter is long over due! Let me tell you a little bit about myself. My name is Jennifer Saunders, a 27 year old college student, and I work part-time at a local mall. I am a cart park attendant at Chris-Town Mall. I ask customers if they would like to park their valuables in a secured area while they shop, eat, get their vehicles, etc. This is a free service provided by the mall. I have been working at the mall for eleven months now.

As far as my education goes, I am currently attending Phoenix Community College this semester. I will get my degree in General Studies (A.G.S) in December '96 or May '97 - I'm shooting for December '96. I want to work in a hospital as a dietitian's assistant. I decided to go into this field due to the work experience program that I went through in my senior year in high school. I worked one on one with a dietitian at a local hospital for seven months. I learned how to do the 10-key, take diet histories of patients, Xeroxing copies of menu planners, and answering phones.

My grandmother was a diabetic for twenty years. During the last couple years of her life, I helped her with medication, meals, etc. I finally decided at that time what I wanted to do with my future. I can say she was an inspiration to me.

School has always been a challenge for me. I started going to school at three years of age. I was in and out of special education classes from preschool to high school. In college, some courses have been more of a challenge than others. I can actually say that I am an average student. I am currently carrying seven credit hours and holding a 2.5 GPA. I will transfer to Arizona State University or Ottawa University in January 1997 majoring in Human Services.

I am the eldest of four children of Patrick and Phoebe Saunders. I was born on July 4, 1968, in Phoenix, Arizona. I was a healthy baby for the first few days of my life. My mother and father were concerned about me because I was not nursing, sucking, etc. My parents admitted me into the hospital at eleven days old. I was flown to Denver to receive the proper treatment for maple syrup urine disease. I faced death six times, aspirated, and had two cardiac arrests. As a result, I have a very mild case of cerebral palsy due to the lack of oxygen to the brain.

The cerebral palsy was more apparent when I was a child than in my adolescent years. I received physical and occupational therapy from an early age until my sophomore year in high school. I never liked physical education until my first year in college, in 1987. Physical education was a challenge for me. I felt that the other children would laugh and tease me. There were a lot of my peers that did tease me and it did hurt me! It wasn't until my high school years that I felt accepted for who I was. I finally learned that those individuals who discriminate against me to this day have the problem, I don't.

I have overcome a lot of obstacles in the past twenty-seven years through family, friends, and my faith in the Lord. I believe that following your diet properly plays a major role in the lives of children with MSUD and their families. Diet control plays an important role in the way an MSUD child behaves.

Behavioral patterns can affect the MSUD child throughout his/her adulthood. Reasoning and proper judgement are the two major areas affecting behavioral patterns. This is why it is important for MSUD patients to watch their diets very closely. Do not feel embarrassed to eat in front of your peers. Tell your peers that your body has a low tolerance for certain foods, liquids, etc.

I have learned a lot from doctors, my parents and from attending MSUD Symposiums. My overall health is very good and I am working out on a daily basis by walking, lifting weights (light), playing volleyball, etc. I have not been seriously ill in 16 years. The only thing is that I've had two wisdom teeth pulled in January, 1996, and I did well. I'll be getting the other two extracted in a few months.

I have a younger sibling, Julia, who has MSUD, too. She was diagnosed at three days of age and is a graduate of Grand Canyon University, 1993 (Communications Major). She is currently working full time. My youngest sister and brother, Jacquelyn and Patrick (Packy) do not have the disorder and yet, they could be carriers. Hopefully, they are not.
I feel that individuals who have MSUD can live a normal life and succeed in whatever they do. May the Lord richly bless all of your lives and I look forward to seeing you at the Symposium in 1998.

***- Sincerely, Julia Saunders***

# FAMILY HISTORY - VERNA MARTIN

**Details**

Written by Lawrence & Malinda Martin

Published: 20 July 2009

**Verna is the daughter of Lawrence and Malinda Martin from Versailles, Missouri. They attended their first Symposium in '94 in nearby Columbia several months after Verna was born. Malinda handed me this history at the our '96 Symposium. Verna has the Mennonite classic mutation of MSUD.**

March 3, 1994, dawned bright and clear - a pure reflection of the joy and happiness that filled our hearts as we welcomed the birth of our second daughter Verna. She weighed 9 lb. 2 oz. and was 22 in. long. She had a head full of black hair. A dream come true.

As we rejoiced, and thanked God, the Almighty Creator, for again having blessed us with the privilege and responsibility of caring and loving another one of his children. Little did we realize the responsibilities, cares and many anxious hours that awaited us in the coming days and months.

Verna was very contented and fed really well the first day. By the third day, she got more sleepy. We tried a lot of things to wake her up enough to feed. By the fourth day, she seemed somewhat irritable-like, just busting out of sleep with a rather shrill cry, and showing some slight signs of arching her back. Her feedings were very short. All of these signs were new to us. It did not alarm us enough to take her to a doctor.

We called our midwife several times, who was unfortunately not familiar with MSUD. But she did advise us to take her to a doctor if our instinct led us to. But as for herself, she felt that Verna was just a very sleepy baby and it would probably take a few more days for her to become more awake and active.

On the sixth day she gave a very nice smile, giving us some hope and encouragement. But we often had a worried feeling. On the ninth day, we had some visitors who thought her cry sounded like a sick puppy. All this time she became less active and would arch her back when we tried to feed her. She only took a few swallows, then she turned blue from choking. She would also take her little fists and push herself away, almost like she knew she wasn't getting the proper formula.

During the night of the ninth day, she awoke almost every half hour with a very scared, shrill cry. Even though she awoke often, she only nursed once during that night.

Saturday, March 12, was the tenth day of Verna's life. Our midwife came to give Verna her check-up. She was rather alarmed when she saw how lethargic she had become by this time. It seemed as though she was blind, as she would not respond with her eyes. She also thought it seemed like Verna had a neurological problem, since she seemed so lifeless. She therefore made an appointment with a neurologist at the University Hospital in Columbia for Monday.

But much to our relief, a higher hand took over, and through the grapevine word was spread through the community about Verna's conditions. A very anxious mother called - the mother of an MSUD child in the community. She very strongly advised that we see a doctor immediately, even though it was already 8:30 Saturday evening. She told us to smell Verna's diaper. We compared the smell with our other daughter's diaper and there was an unmistakable difference. Her husband very graciously called their doctor and made an appointment with him to meet us in the emergency room yet that night.

The stark news struck us at first. But we were relieved to know she wasn't blind, and now we knew what was wrong with our baby. It was a relief to hand her over to the professionals to take care of her.

We met Dr. Richard Hillman and Julie Grasela (the dietitian) at the University Hospital in Columbia, MO, where (Verna) was diagnosed with MSUD and put on an IV. She was also started on formula. She would not take her bottle, so she was fed through an NG tube. Her leucine levels were at 4186 micromols/liter *(55 mg/dl)*.

After being in the hospital 10 days, she was brought home, where we slowly tried to learn how to care for our baby. At first it seemed like we would never learn. But by taking one day at a time, with the encouragement and help from friends, neighbors and others, and most of all, much strength and guidance from above, we slowly made progress.

She had ups and downs as we were trying to keep her amino acids in line. It seemed the first year her levels would fluctuate an awful lot. When she was 3 months old, she became very deficient in isoleucine and valine and was hospitalized for 6 days until her levels returned to where they should be.

She had her share of the flu. Some times we had to put the NG tube down to feed her till she was over her flu. But she had a lot of good days. She eats well and takes her formula well when her health and levels are in line.

Verna is now 2 years old, and is a very healthy girl. She is truly a blessing to our home, and has brought much sunshine into our lives. We have often been strengthened by her will power and courage and many smiles, even when she is coping with a flu that has elevated her levels. Even though providing for the many needs (physical and material) has been a challenge, and sometimes almost seemed impossible for a child with MSUD, we believe we would all admit that providing the soul with its spiritual needs and being a good example is still the greatest challenge. And as with all other needs, is only possible with help from God.

May we be strengthened spiritually, and drawn closer to our God, as we willing take care, to the best of our knowledge, of God's special children. May God bless you all,

***- The Lawrence & Malinda Martin Family (Lucille 4, Verna 2, and Lavern 1)***

# MANAGEMENT TIPS

**Details**

Written by Joyce Brubacher

Published: 20 July 2009

**Urine for DNPH testing:** To obtain a urine specimen from an infants diaper, put several cotton balls or sterile gauzes around the child's penis or vagina and cover with a 2 to 3 inch piece of plastic wrap. Put on diaper. When wet, squeeze water from cotton balls or gauze to test with DNPH. No more sore bottoms from urine bags.

**Medicine for vomiting:** Several parents in PA expressed their appreciation for the medicine Zofran (Odansetron) prescribed for vomiting during an illness. The medicine is often used for chemotherapy patients and post surgery patients. It controlled the vomiting in the children with MSUD so they could continue taking their formula and liquids. This is a very expensive prescription drug. If your child vomits easily when sick and often needs to be hospitalized, you may want to ask your doctor about using it.

**Toothpaste for finicky children:** A mother had problems getting her children to brush their teeth until she discovered Tom's of Maine natural toothpaste with fluoride. It comes in delicious mint, fruit and other flavors including Silly Strawberry for children. It is available in health food stores.

# CONGRATULATIONS TIFFANY!

**Details**

Written by Joyce Brubacher

Published: 20 July 2009

We sincerely commend Tiffany Gant who graduated from Providence Catholic High School this spring. She received a NAACP Scholarship and is attending a Jr. College this winter. She is also working part time at Montgomery Wards as a clerk. Tiffany, with classic MSUD, has done very well. Her parents, Terry and Linda, are very proud of her, and we extend a hearty congratulations and wish her God's blessings as she continues her studies.