VOLUME 12-2

Fall/Winter, December 1994

WHAT THE FAMILIES HAD TO SAY

Details

Written by Joyce Brubacher

Published: 17 July 2009

The symposium was an interesting meeting for us. We did not realize there are so many families with MSUD children. Making new friends and meeting old ones was the highlight of the Symposium for us. The professional talks were very informative, too. We had an enjoyable trip. - *A father*

We enjoyed being at the Symposium. It was very well organized and the facility was ideal. We were encouraged to see other families with MSUD children have a normal family life. - *A mother*

We treasure the time spent with other MSUD parents and friends. The speakers did their duty to educate us about new products and ideas. It was refreshing to be reminded again of our parental duties in caring for our children. - *A mother*

We had a very enjoyable time visiting with the parents once again. It was well organized and each speaker was interesting. We need all the encouragement and advice we can get to care for these children. We also thought the motel was a great place to have it. - *A mother*

It was a well-organized conference with very good speakers. The facility was great. We could not ask for better accommodations and they had a pool! Next time I wish they would request notes from the speakers and distribute them to the group so we can follow along and keep the notes for later use. - *Mary Ann Peters*

I felt really welcomed at the Symposium and was very impressed with Dr. Holmes Morton. I found the information on glutamine and isoleucine supplementation fascinating. I took a lot

home from this conference that I have been able to put to good use in caring for our son. I learned not to view the "formula" as such, but as a normal diet requirement that has now been renamed by our seven year old as Mike's Maxi-Milk. He was getting upset that people would think he was a baby if I called it formula. - *Anna Toth*

It was interesting to be there and realize we are not alone in caring for these children. We enjoyed the speakers, especially Neil Buist. He made us realize how far they have come in treating these children. We have many good memories. - *A mother*

Even though the 1994 Symposium was a first for us, it will never be the last if God permits. We so thoroughly enjoyed it. We enjoyed the speakers and gaining new insights and knowledge about MSUD, but most of all, it was a thrill to see someone else in our boat. It was so "weird" not to have to explain what MSUD was to everyone we met. We felt such a kindredness to everybody. It was like a big family. We were so uplifted to see so many kids that were doing so well. All of the children were beautiful.
We'll never forget meeting our first new friend with MSUD. After we arrived at the hotel, we were hoping we were at the right place. We decided to take a swim. Sherwood started talking to a little girl who was swimming and playing with her brother. When he found out she had classic MSUD, he and I were overwhelmed. She was so perfect. This was Leanna Peters. Then, of course, we met her mother, Mary Ann who knew all about MSUD. From there on, we kept being overwhelmed. It was a wonderful, fantastic experience and we recommend everyone participate at the 1996 Symposium if at all possible. It gives you new hope to come home too. Thanks again for all of the hard work. Hope to see you in Ohio. - *Sherwood & Sonya Webb*

I could not do justice in a Newsletter to the material presented at the Symposium. There was so much of value and I hope many of you will take advantage of the videos. One mother arrived home from the meeting to find her daughter ill. She told me the information she had heard was extremely valuable in helping her provide adequate treatment for her daughter. She thinks the outcome could have otherwise been disastrous. Others have testified to the improvement in their children since they have a better understanding of the diet and its role in MSUD.

For most families, sharing on a family to family level is so refreshing, as attested to in the personal notes above. And there are always times of humor; one being the swimming pool incident.

A father attending the Symposium was supervising his child in the pool when he began a conversation with another father staying at the motel. He mentioned that many of the children in the pool have maple syrup urine disease. The other father quickly motioned to each of his children to come out of the pool and they left. The next day or maybe that night, I am not sure, the pool was loaded with chlorine tablets. Several fathers threw some out but enough was left to burn the eyes of the swimmers. It wasn't until the meeting was over that some of the parents began to

wonder. Did the "urine disease" have anything to do with the highly chlorinated pool?

NUTRITION PANEL

Details

Written by Joyce Brubacher

Published: 17 July 2009

Panel members: Singh, Buist, Trahms & Grasela

Q. Is it OK to freeze the MSUD formula after it is prepared?
A. Freezing the formula can break down the fat emulsion. It is not necessarily a good idea, but there is no chemical problem with freezing. Freezing will make the formula difficult to mix.

Q. How long will mixed, prepared formula last?
A. It is recommended that the prepared formula be treated like cow's milk. Bacteria formation is the biggest concern if it is not refrigerated. Within four to eight hours, bacterial growth can start.

Q. What are some high calorie, low-protein foods?
A. Baking mixes are the best. What is best is what the child will eat. The lower fat products are easier on the stomach.

Q. Is it OK to give persons with MSUD aspartame (Nutrasweet)?
A. Yes, only persons with PKU should avoid aspartame. Most persons with MSUD need calories and do not need this product.

Q. Why do computer programs vary on the protein content in various foods?
A. The database is old and needs to be updated.

Q. How often should you check DNPH and have blood work performed?
A. When there is a rapid growth period, DNPH and blood work must be done more frequently.

A CLOSER LOOK AT HOME MONITORING TECHNIQUES

Details

Written by Joyce Brubacher

Published: 17 July 2009

Presented by Richard Hillman, M.D. - Columbia, Missouri

DNPH is a test that measures keto acids, ketones and acetones. Normal newborns do not excrete ketones; their brains are using the ketones produced by the body. DNPH will test positive when the BCAAs are elevated in a child with MSUD. It is a good test to use for home monitoring.

Use an equal amount of urine and reagent. When the reading is negative, it looks very much like the reagent. If the reading is cloudy, the individual with MSUD has elevated levels. If the reading shows a precipitate (settlement on the bottom of the test tube), illness is definitely indicated. The solution gets more orange with time after it is mixed. It is best to read the test right after the urine is added.

DNPH is a strong acid that will eat through material. Refrigerating the solution in a dark bottle will extend the shelf life, but the solution should warm up before it is used to catch a slightly positive reading. It is possible to get a false positive with old DNPH. Do a test on a person who does not have MSUD to check it.

Newborns often produce a slightly cloudy reading that is false. A slightly cloudy solution can be falsely positive due to the gels in the new disposable diapers. Also the urine of babies is often mixed with stool. In addition, two antibiotic prescriptions for newborns cause false positive readings.

Vaginal discharge can also cause false positive readings in older girls. A clean catch urine sample must be taken to avoid these false readings. DNPH is very important for testing older children who are harder to judge if their levels are off. One sign of high levels is deterioration in handwriting.

Ketosticks measure ketones that are not from MSUD, but produced from fat breakdown, such as when a person is very ill or has fasted. DNPH is much more sensitive.

Dr. Hillman demonstrated the DNPH by using the urine from a baby with MSUD, who was sick at the time. The reaction was very positive with an immediate, very cloudy (opaque) reaction.

WEST COAST EXPERIENCE WITH MSUD

Details

Written by Joyce Brubacher

Published: 17 July 2009

Presented by Neil Buist, M.D. - Portland, Oregon

There are 130 metabolic diseases treated in Oregon. Oregon was the first State to set up screening for MSUD. Dr. Buist showed slides including the first infant with MSUD to be identified through state screening (Monte Brubacher, son of Wayne and Joyce, 3/12/65-12/4/74).

Certain amino acids, vitamins and trace elements cause the bad taste and smell in most formulas. Dr. Buist and the Oregon Clinic developed a good tasting MSUD formula that Dr. Buist brought along to the Symposium. In it they eliminated some nonessential amino acids and adjusted the amounts of others. It is similar to the new PKU formula Periflex, developed at the Clinic and distributed by S H S North America (new name for Scientific Hospital Supplies). The adults and children sampled the new MSUD formula.*****

There is the possibility of using the amino acids to make foods that look and taste like their counterparts but do not contain the BCAA. However, amino acids cannot be baked. They are best used in spreads. There is research being done on a protein free cheese. Dr. Buist's dream is to make a BCAA-free hot dog someday!

Massachusetts has a law that requires manufacturers to add a medical label to medical foods. This makes it easier to get insurance to pay for the foods.

Phenylalanine is a certain percentage of protein but leucine amounts vary in different foods. A very general rule for figuring leucine is to double the phenylalanine amount in a food.

Questions from the audience

Q. Is MSUD included in the Oregon health plan?
A. MSUD is very well treated in the health plan, since there is a treatment available, and death is the result if it is not treated

Q. What about the National health care plan?
A. Dr. Buist fears that the metabolic diseases will be discriminated against, because they are of a minority group. (Ted Kennedy's health care plan includes language for coverage of medical foods.)

*** I was skeptical when Dr. Buist handed me a cup of liquid and said, "Taste it." Hm-m-m, it looked good and yes, it even smelled good. I took a tiny sip and immediately took another drink. To me it tasted like orange sherbet. And believe it or not÷no aftertaste! All of the other formulas make me gag at the smell and the taste is worse than the smell. This new**

formula enthused the parents but not all the children.

Many children with MSUD are skeptical of anything that looks a little different or is "good for them." Each child was served a cup of the formula during one of their break periods in the activity room. They were not told it was formula and their reactions were video taped and shown later to the parents.

It was quite amusing to see them look at this strange looking drink. Some drank it down without the least suspicion÷especially the siblings of the children with MSUD. Some would hesitate, take a small sip, set it down and act so undecided. Others with MSUD refused to touch it. The teenagers and young adults liked it. Shayla thinks it would be great.

One girl with MSUD wouldn't taste the drink, but her sister asked for refills during the children's break and later was filling her cup from the pitcher in the conference room. I think the new formula would be great for those who dislike their current formula and for the newly diagnosed.

Dr. Buist says it should be no more expensive than the current formulas. It is not the ingredients that are expensive, but the processing. It may be more expensive if the whole family wants to drink it! Imagine having formula in the refrigerator that doesn't create a catastrophe if a friend begins to drink it by mistake. Hurry Dr. Buist, hurry, get it on the market.

THERAPY DILEMMAS OF THE OLDER CHILD WITH MSUD

Details

Written by Joyce Brubacher

Published: 17 July 2009

Presented by Holmes Morton, M.D. - Strasburg, Pennsylvania

Most of what is written about the care of children with MSUD stops at one month of age. There is little consensus on proper treatment of the child with MSUD. Dr. Morton is interested in writing a standard treatment protocol manual for doctors.

Protein tolerance is about 20 mg/kg from two years old until adolescence. It changes with the rate of growth; there is an increased need for leucine during growth spurts.

With increased physical activity, protein is broken down, causing fatigue, weakness and ataxia. This is due to the muscles depleting glycogen stores and breaking down stored protein for energy. It is best to use high fat foods prior or during strenuous activity to provide energy.

Burns are catabolic injuries and sunburns can also cause this breakdown of protein if adequate calories are not provided. Fasting, even overnight, causes protein to break down at the rate of 1gm/kg/day. All of this must be considered during sick days. During sickness the child is not growing and protein needs decrease. It is important to restrict activity during illnesses, provide adequate calories and make sure there are no long fast periods. Wake the child every few hours to give formula/calories.

The anesthesia used for surgery doesn't cause any changes in BCAA levels but should not be a kind that causes vomiting or paralysis of the intestine. In orthopedic surgeries there is the danger of immobilization. Not eating for 24 hours after surgery can start catabolism (the break down of protein). Head injuries and surgeries, and some medications used with head surgeries, cause extreme catabolic problems. In postoperative care, glucose and insulin IVs should be added to decrease leucine levels rapidly. Glucose suppresses the growth hormone.

Cerebral edema is not clearly understood and cannot be predicted by leucine levels. Dr. Morton has successfully used Phenobarbital, Manatol and glutamine to treat cerebral edema.

Glutamine is highly concentrated in muscle cells and inhibits muscle degradation in high concentrations (the cells don't grow unless there is a high concentration of glutamine). It is important for the immune system. Dr. Morton uses 200 to 500 mg/kg/day of glutamine when a patient with MSUD is sick. Sick people don't make glutamine as well and individuals with MSUD especially make less glutamine. They only get about 1/2 of the glutamine most people get in their diets. There is 4 mg/kg of glutamine in the Ketonix formulas; that is 1/2 the normal amount. Other formulas have even less.

Pancreatitis is recurring more often in patients with MSUD. Be on the lookout for pancreatitis, hypoglycemia and diabetes. When someone with MSUD exhibits sweatiness, a feeling of unease, and "the shakes," check the blood sugars. Dr. Morton does not believe that the MSUD formula necessarily causes hypoglycemia. Problems with thirst may indicate diabetes. Check for sugar in the urine.

Questions from the audience

Q. Dr. Morton, do you routinely supplement the MSUD diet with glutamine?
A. If there is great fatigue, poor appetite, or lack of energy, Dr. Morton will consider it. Glutamine will not hurt; it is available in health stores.

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UPDATE OF NUTRITION MANAGEMENT OF MSUD

Details

Written by Rani Singh, PhD.

Published: 17 July 2009
Submitted by: Rani Singh, PhD. - Atlanta, Georgia

MSUD is a genetic disorder. Children with this disorder cannot use the BCAA (branched-chain amino acids) in the normal way. The outcome in these children is affected by several factors. These include age of diagnosis, degree of enzyme impairment, time at which diet therapy is begun and adequacy of management. Management requires an aggressive protocol to reduce plasma BCAA to avoid central nervous system changes. Goals of long-term nutrition support are:

a) Normal plasma amino acid concentrations
b) Urine free of branched-chain keto acids (BCKA)
c) Normal growth, development and status.

Nutritional management requires initiation of nutrition support immediately with BCAA free medical foods (formula). Depending on clinical status, patients are fed by nipple, nasogastric (N-G) tube, intravenous infusion (IV), or a combination of these methods. When plasma leucine reaches the upper limit of treatment range, leucine is added back gradually using infant formula

or whole cow's milk depending on the child's age.

Over restriction or imbalance of BCAAs has adverse effects. These include anemia, desquamation (shedding of the skin) and failure to thrive. Free isoleucine and valine can be used to correct some imbalances. It is very important to provide protein-free sources to meet energy requirements. In addition 100 to 500 mg of oral thiamine per day may be provided for three months to evaluate the response. Guidelines used for fluid requirements in treating these children include:

- Less than one month of age:100-150 ml/kg/day
- 0-10 kg in weight:100ml/kg/day
- 11-20 kg in weight:1000ml+50ml/each kg over 10kg
- Over 20kg in weight:1500ml+20ml/each kg over 20kg

It was emphasized that energy requirements during acute illness/surgery increase. Therefore aggressive nutrition support becomes necessary to prevent catabolism which could be caused by poor oral intake or the stress of infection itself. Suggested nutrition protocol during illness:

1. As soon as the child shows any signs of being sick, natural protein should be reduced by half or eliminated.
2. Adjust energy intake (kilo-calories) through medical food (protein-free) to at least maintenance levels. Try to add 10-20% above maintenance levels.
3. All fluid needs could be provided through the medical food mixture.
4. Give feedings and drinks every 1 to 2 hours in small amounts to children less than 5 years old and older than three.
5. Continue all vitamins and medications at the same dosage.
6. Minimum volume intake of 75% of prescribed intake for two consecutive feedings and 85% of prescribed intake during 24 hours should be acceptable.
7. If decreased intake continues for 24 to 48 hours, make an effort to maintain intake by nasograstic tube using enteral pump. The oral regimen is preferable to intravenous therapy.
8. If nausea or vomiting persists and the gut cannot be used, peripheral venous alimentation can be used for patients whose needs are likely to be short term (2 to 5 days). Central hyperalimentation should be implemented for patients requiring long term support.

In conclusion, immediate adequate energy and fluid intake become important during acute episodes.

ELSAS RESEARCH UPDATE AND UNDERSTANDING THE ROLE OF FORMULA IN TREATING THE DISEASE

Details

Written by Joyce Brubacher

Published: 17 July 2009

Presented by Richard Hillman, M.D. - Columbus, Missouri

Dr. Hillman opened with the statement, "We want you to know more than your doctor." He reported on a research project by Dr. Elsas from Atlanta, Georgia. At the Toronto Symposium, Dr. Elsas had a number of children with MSUD drink a small amount of leucine and then blow into a balloon. He measured the carbon dioxide expired. (*See the Dec. '92 issue of the Newsletter*.)

The children had a very low amount of leucine in their breath compared to adults without MSUD. Unfortunately, carriers of the MSUD gene could not be readily identified, because their levels were too close to normal. The enzyme level correlated with the leucine level in the breath, but the test is not practical to use except to possibly help establish early dietary treatment. It seems more important to adjust the diet to the individual than to the level of enzyme activity.

Since the enzyme block is in the conversion to keto acids, the levels of keto acids seem to be more important, but it is easier to measure BCAA (branched-chain amino acids). The BCAA need to be turned back into their keto acids so the body can use them or eliminate them via the urine. The amino acids, alanine and glutamine, add ammonia to the system helping to turn keto acids back into amino acids. MSUD formulas should be supplemented with these two amino acids.

Formula is therapeutic and should be given when sick. It can be given with an NG tube, IV, or central line (which can be used when administering TPN). Dialysis removes BCAA but does not remove their keto acids very well.

Questions from the audience

Q. Why isn't alanine and glutamine automatically added to the formula for all persons with MSUD?
A. Dr. Hillman recommends it. Glutamine is unstable and has a shelf life of 18 months. Glutamine loses one ammonia, causing a bad smell (thus the comment that it tastes bad). However, when refrigerated, it is kept more stable and doesn't have any flavor.

Q. Are doctors talking to each other about the different techniques for treating and managing MSUD?
A. Somewhat, but not very well.

PANEL: ASK THE DOCTOR

Details

Written by Joyce Brubacher

Published: 17 July 2009

Panel Members: Drs. Hillman, Berry, Morton, Grange

Q. What causes the stomach cramps?
A. The formula is similar to predigested (highly concentrated) formula; it can cause irritation in the stomach and intestines, particularly when the formula is thick.

Q. Why are valine and isoleucine used when MSUD people are sick?
A. When there is a high leucine level, more of the other amino acids are required to make protein (use the excess leucine) to lower the leucine level. In addition, sufficient calories are necessary for protein synthesis. The amounts of additional valine and isoleucine vary depending on the patients, circumstances, etc. (A good assumption is to try to make the valine and isoleucine levels as close to normal as possible.)

Q. When leucine levels are elevated, where does the leucine go and what damage does it cause?
A. The leucine goes to the next metabolic step, forming the keto acids, which are present in the urine. Leucine inhibits phenylalanine in the brain. However, the keto acids are the primary poison. These acids interfere with growth, causing malnutrition. They also draw water into the cells.

Q. Why are there different management techniques (counting leucine vs. counting protein)?
A. Persons with MSUD can be normal when on well-controlled diets. However, if these same people are on poor diets, they will perform poorly. Children with leucine levels 5 to 10 mg./dl may behave like children with Attention Deficit Disorder. Dr. Morton feels that most problems experienced when levels are high can be reversed if the people are kept on well controlled diets. He likes to see leucine levels at 2 to 3 mg./ dl. People kept at 500-1000 micromols/Liter of leucine do not do as well as people under 500 (behavior is worse, etc.). In addition, persons with MSUD seem to have better recuperative abilities (such as from a coma) than most people.

Q. Does isoleucine keep leucine and valine stable?
A. All three are needed in the correct balance and available at the right time of protein synthesis to function correctly. However, leucine seems to be the only one that causes reactions. But you cannot regulate just leucine. All three must be balanced.

Q. Do all three amino acids cause ataxia?
A. If the valine is high, there are no symptoms. If isoleucine or leucine is high, ataxia will occur.

Q. Does it matter if a person with MSUD fasts or does not eat regularly?
A. Yes! Individuals with MSUD can go a normal time between meals, but it is important to regularly take in carbohydrates. The BCAAs have a lot to do with appetite control.

DNA REVIEW AND UPDATE

Details

Written by Joyce Brubacher

Published: 17 July 2009

Presented by Kathy Grange, M.D. - Columbia, MO

Chromosomes are made of genes and contain DNA. The location of the genes is random, and the MSUD genes are located on many different chromosomes. Mutations (inborn errors) occur when there are deletions of base pairs, insertions, or wrong combinations. In a mutation, there may be no enzyme where there should be, or an abnormal enzyme. The abnormalities are what usually causes metabolic disorders.

Mennonites are homozygous (the same mutation exists on both genes). Non-Mennonites have a good probability of being compound heterozygous, one mutation from the mother and one from the father. MSUD is more complicated than other genetic disorders due to so many mutations.

Gene therapy requires locating and isolating the mutated gene. Family members can be tested for carrier status if the mutation is known. The mutation in Mennonites is known.

There are currently two methods of gene therapy÷injection of "fixed" genes into cells and viral mediated transfers. Direct injection is tricky and requires the removal of tissue from the source, injection and growth in a cell culture, and placement back into the source. With viral mediated transfers, it is necessary to prevent the virus from acting on its own (for example, a virus can insert itself in the wrong place and cause problems).

Researchers have successfully done viral mediated transfers in a beaker for MSUD. The next step is to try it on cows known to have MSUD. There are still questions as to how long the enzymes will remain active once the person has been "cured." It may be long term, or there may be periodic doses required, or it may be a supplement to diet.

FEED THE CHILD, TREAT THE DISEASE

Details

Written by Joyce Brubacher

Published: 17 July 2009
Presented by Helen Berry, M.D. - Cincinnati, Ohio

In MSUD, there is a metabolic block at the second step of the metabolic process. Babies start out OK, but, if untreated, they will begin with hyperactivity, convulsions, coma and eventually death. There is a rapid progression when the BCAA (branched-chain amino acids) levels become elevated. Ten to fifteen days may be too late to correct any mental problems that occur from the elevated levels.

MSUD is difficult to treat because there are three amino acids to keep balanced instead of just one as in PKU. In the early years, the doctors mixed the child's formula using the required BCAA. At first methionine was also restricted in the diet.

The first child treated was born to a California family in June '59. (A sibling died earlier from MSUD.) It was first thought she had colic but they noticed the sweet odor on the 11th day. She was taken to New York where they confirmed the diagnosis. When the child was around 22 months, blood testing procedures had improved enough to determine a difference between methionine and alloisoleucine. So methionine was added back to the diet. She stayed at the hospital under the care of Dr. Snyderman for four years. She got sick every time they took her home and was not growing well. She was 4¸ years old before she passed the third percentile.

This child moved to OH in '72 and became Dr. Berry's patient. In '77 she joined a group home for women. At 18, her IQ was 64, and she had the social skills of a 12 year old. She eventually got a job at a fast food restaurant. When she started gaining weight, it was suspected that she was cheating on her diet. At 19 years an EEG showed a generalized convulsive disorder. In 1990, she was admitted to a mental institute for confusion. She was placed on her diet and improved. As far as it is known, she still may be cheating, but is doing well. It is important to train physicians to deal with adults with MSUD.

Another child, born in Chicago in 1976, eventually became Dr. Berry's patient. She was hospitalized at one week of age. Although lethargic and not eating, she showed no elevated BCAA levels. She was later released with a diagnosis of a mental episode. In the hospital, she had been on a protein-free IV, so her metabolic condition stabilized. She was hospitalized again at four months of age with projectile vomiting. A nurse noticed the sweet odor leading to a correct diagnosis and treatment for MSUD.

(At this time Scientific Hospital Supplies in England was making MSUD Aid. It was difficult to import as customs raised the fees from 5 to 25%. They said it was a petroleum product.)

This second girl and her family moved to Dayton, OH. A pediatrician cared for her at first and then turned her over to the clinic. In 1978 she became Dr. Berry's responsibility. She had had poor growth and many hospitalizations.

Dr. Berry noted leucine plasma levels were reduced and growth improved when valine was added to the diet. She made a DNPH kit for the mother with the following instructions:

- A trace precipitate within 60 sec. and negative ketones = no treatment required.

- A slight precipitate and negative ketones = watch the patient.
- A moderate precipitate within 30 sec. and positive or negative ketones = increase fluids, decrease BCAA.
- A strong precipitate within 10 sec. and positive ketones = eliminate BCAA from diet; repeat test every 12 hours.

They did a series of studies to see why she had so many hospitalizations. It was determined to be very important to monitor **all three** amino acids. On a fast, BCAA levels started rising after six days. The lowest level was an hour after each meal and the highest was after an overnight fast. The following guidelines for plasma concentrations were used for treating the patient.

- Leucine: 20 mg/kg or 180-70 micromols/l
- Isoleucine: 12 mg/kg or 70-280 micromols/l
- Valine: 14 mg/kg or 200-800 micromols/l

The girl now has isoleucine and valine added to her formula to keep her levels stable. So far, there have been no more acidotic episodes.

Questions from the audience:

Q. Originally, a girl had her diet supplemented with isoleucine. Now she is 17 and there are no problems with low isoleucine. Why?
A. Dr. Berry feels that puberty may affect MSUD people to the point of improving the tolerance. However, another mother in the audience said her daughter's tolerance is worse since she began puberty.

Q. What would a low leucine level be?
A. 150 to 200 micromols/liter of leucine.

Q. What causes hypoglycemic conditions?
A. High leucine levels seem to affect this condition, but more work is needed before it is fully understood. We need studies to see the relationship between amino acids and insulin.

FAMILY HISTORIES - SAD NEWS

Details

Written by Joyce Brubacher

Published: 17 July 2009

This summer we received word of the death of Scott C. Foster. Scott was born on Aug. 17, 1971 and died on May 9, 1994. At 22, Scott held a full time job and was a busy young man.

He was doing so well no one realized how quickly things could change. He died after a short illness. Scott had classic MSUD.

The Foster family has established the Scott C. Foster Memorial Fund. The following is taken from a letter describing the Fund.

"The loss of Scott Foster is something that will be felt and remembered by many, and that is particularly comforting to the Foster Family during this time. Those of you who knew Scott will remember a very special and caring person.

"While it is good the disease is so rare, this also creates a problem of limited research funds and attention to find a cure. Many in the medical profession expect with proper funding that hopefully a cure will be found in less than a decade.

"At this time, the Foster family and friends have decided that the best remembrance of Scott will be to dedicate time and effort toward fighting the disease that took Scott and leaves many children at risk. One of these children at risk is Scott's beautiful younger sister, Katie, who is now 11 years old, and also has MSUD.

"To that end, the Foster family has created the Scott C. Foster Memorial Fund that will be dedicated to raising money to fight this disease. All funds received toward this fight will be channeled to a research group at the Massachusetts General Hospital who will use the funds for genetic research."

I understand the Fosters are very busy with their fund raising activities. May the Lord bless their efforts. It is a wonderfully healing way to work through our sorrow by putting our efforts into helping others. We extend our sincere sympathy to the Foster family.

FAMILY HISTORIES - LETTER FROM ANDRIA MERRILL

Details

Written by Andria Merrill

Published: 17 July 2009

Some of you may remember Ron and Andria Merrill who attended the Colorado and Pennsylvania Symposiums. We were disappointed when issues of the Newsletter were returned with no forwarding address. It is good to welcome them back into the organization and print this updated information on Ryan.

Dear MSUD Family,

It has been a long time since we have heard from you. We lost track of the Newsletter through all our moves over the years. Because it has been so long, I thought you might like a brush up on Ryan's life over the past years.

Ryan was born on June 12, 1982 in Bountiful, Utah-the only child with MSUD in Utah. I was 18 years old and Ron, my husband, was 21. What a shock! He wasn't diagnosed until he was six weeks old. We had him in Primary Children's Medical Center (PCMC) two of his first six weeks.

During the second admission after testing for everything possible, they ran the test for MSUD. It came back positive. We didn't understand the disease at all.

Ryan had symptoms similar to your children-rejecting formula (Similac), constantly crying, weight loss and dehydration. He wouldn't open his eyes and seemed in constant pain. After he was diagnosed, we spent approximately three months in PCMC.

During this time we met Metabolic Specialist, Dr. Claire Leonard, who works out of the University of Utah Medical Center. During those three months with her, we learned how to take care of Ryan. I was so overwhelmed. I think I aged about 10 years in three months.

When we took him home, it was one of the greatest moments in my life. I didn't have to share my baby with a nurse or doctor. We could finally start the bonding between mother and infant. The Lord truly blessed us. Our son was alive and doing well.

Of course, over the next three years we had our many visits to PCMC. Most visits were for high protein levels or low glucose levels. There were a few times we almost didn't bring Ryan home with us. Ryan has classic MSUD with the complication of low glucose. When Ryan becomes ill, we often end up at PCMC-usually for only a day.

As Ryan grows older, he seems to take a longer amount of time to bounce back. We now have spent up to four days at PCMC. No matter how old he gets, it doesn't get easier. I don't ever leave his side. I want to be there if the Lord calls him home.

Ryan attends a "normal" school. He mainstreams 50% of the day in his normal 6th grade class. The other 50% is spent in a special class for the disabled. Ryan is 12 years old. He weighs 52 pounds and is 50 inches tall. He loves to play basketball and very rarely misses a UT Jazz game. He had the opportunity to go to the Jazz games and meet some of the players. Due to some brain damage at birth, he has spastic ligaments and hamstrings, but is walking on his own after having approximately seven surgeries.

Ryan has two healthy sisters that he loves to torment. We know the day will come when Ryan will return to his Heavenly Father. We pray that the Lord will continue to trust us to raise one of his special children, for he has touched our hearts forever.

- All our love, Andria & Ron Merrill

FAMILY HISTORIES - SOMETHING WRONG WITH GLENN

Details

Written by Daniela Hudson

Published: 17 July 2009

Daniela Hudson sent a letter written on June 24, 1994. It gave an account of her son, Glenn, and his very late diagnosis of MSUD. He has done quite well since he started on the diet.

My name is Daniela Hudson. My husband Glenn and I have two children, Glenn Daniel, 8 years old, and Amanda, 5 years old. Glenn has intermediate MSUD and was diagnosed three years ago at Children's Hospital in St. Louis, Missouri. He was 5 1/2 years old.

I knew from early on that something was not right with Glenn. That was especially clear after my daughter was born and I saw how she developed. It was like night and day. Glenn as a baby was always sleeping and a poor eater. He fell asleep during feeding. Amanda was very alert and very good at feeding. Glenn never learned how to turn over; I taught him how to crawl. Amanda did all those things on her own.

Glenn didn't learn to walk until 18 months and then only with the help of a therapist. Amanda walked at 11 months. Glenn didn't start to talk till three years of age and Amanda started much, much earlier. I also noticed "attacks" or "spells." He couldn't keep his balance, and had slurry speech, glassy eyes and sometimes vomiting. Every time that happened, I took Glenn to the doctor. They either didn't know what was wrong with him or by the time I got there, the "spell" was over. Once a doctor saw it, and did all kinds of tests, but never found anything.

At the age of three, he was hospitalized 10 days for observation. Nothing. At the age of 5 1/2, he had the worst "spell" ever and was transferred to St. Louis Children's Hospital. My husband is in the Army, and we were stationed at Fort Leonard Wood, Missouri at the time.

Glenn's spell started in the afternoon and lasted the whole 2 1/2 hour drive to St. Louis, continuing after we were there. I told them Glenn's history, and they started testing. Within hours I had the news. Indeed Glenn had something with a name and it wasn't all my imagination. To my husband and me, it was a great relief. Glenn was put on a diet of 14 grams of protein from food and on formula called Maxamaid. The formula was quite a challenge. Glenn did not like it at all. The diet was no problem. He never ate dairy products and only had to give up hot dogs and bacon. He didn't give us any struggle with that.

Then came our move to Hawaii. The first few months were hard. We were still getting used to the idea of Glenn having MSUD, his diet, blood tests and starting kindergarten. There

was financial hardship because of the move and hospital bills. But we got through it.

Receiving your MSUD information packet was very helpful. I especially enjoy receiving the MSUD Newsletter. It makes us feel that we're not alone and has lots of information. I also know that I can reach out to someone who knows what I'm talking about.

Glenn has a great dietitian here on the island. She, Robin, decreased Glenn's protein from 14 gm to 10 gm after seeing his blood levels. From measuring his food by cups and spoons, we went to using a gram scale and exchanges. Robin also added solutions to his formula. They made quite an improvement in Glenn's behavior. Robin also gave me some hints on how to prepare formula so Glenn would like it. Instead of water, we use Kool-Aid, and that did the trick.

Glenn also has a great doctor. Dr. Hsia was always there when Glenn was not well. Glenn was hospitalized twice during our three years on the island. The first time it was for headaches that affected his everyday life. A teacher from school had called and told me that Glenn was not himself. Once in the hospital an MRI revealed acute sinusitis.

The second time he had a virus that caused him to become acidotic from fever, vomiting and not being able to eat and drink. Once in the hospital, he got better fast.

After being on the diet and on formula (Ketonex 2) for three years, Glenn is doing very well. In school, Glenn is an average student. He goes to special education part time for reading, writing and math. He does better in a small classroom for those subjects. He goes to speech therapy once a week and is in his regular classroom for everything else.

Glenn just got done playing baseball. It was his first sport. He had fun but it tested his patience when he had to be in the outfield. He liked batting a lot better.

Glenn loves to be outdoors. He also loves to play Nintendo, Sega and cartoons. He gets along with almost everyone.

Glenn and his sister are enjoying the summer and our last few weeks here on the island. We are leaving July 7 to be stationed at Ft. Campbell, Kentucky. First the children and I are going to Germany to visit my family. My husband will set up house and spend the time with his family in Indiana.

I think the support group is really great. We're sorry we couldn't make this year's symposium. We'll be at the next one for sure.

SHARING - A POSITIVE LEARNING EXPERIENCE

Details

Written by Sandy Bulcher

Published: 17 July 2009

After listening to Christine Trahms, the dietitian who spoke at this summer's Symposium, I became motivated to include Jordan (age 5) more in the management of his diet. I started by encouraging Jordan to report what he had for a snack and how much he had eaten while playing at the neighbors. He'd proudly run home and announce "I had 2 pretzel sticks and apple juice."

When I pick him up from preschool, he's eager to report exactly what he's eaten, also. (I still do rely on the supervising adults to report specifics, however.)

While older brother, Tyler, is at school, Jordan and I play school at home. He's learning to read the number of grams of protein from nutrition labels. I place several different cans or boxes of food in front of him and he finds the nutrition label and reads the number of grams of protein. He then decides if he can have "as much as he wants," "a little," or "none" of the food by the amount of protein. He loves to play the "protein game" and I end up emptying the cupboards and drawers before we are done.

We have a lot of fun, and it's been a positive learning experience. It has given his self-esteem a big boost!

SHARING - DIANA AT THE SYMPOSIUM

Details

Written by Diana J. Blum

Published: 17 July 2009

I am writing to tell you that I enjoyed myself at the Symposium. I found the panel especially interesting when I was chosen to participate on the panel. I hope I was able to help or give ideas to parents.

I also found the teen discussion quite informative. For example, I learned that once I am older and married, I can have kids. If my husband is a carrier then our children will have classic MSUD. That is, since Iâm a carrier also. If he is not a carrier then our children will only be carriers and not have the disease. I also learned that there is a cow in Australia that has MSUD, and scientists are doing studies, and might have a cure for this disease. I surely hope they do.

I learned how to deal with MSUD. And finally, I learned how people react to those who have MSUD and how to deal with them. This will come in handy.

For example, I meet other kids in school and we start to talk. When I tell them I have MSUD, they call me a liar. So instead of getting mad, I prove it to them by showing them my medic alert bracelet and information card. When they realize it is true, they think it is weird. But they become my friend because they think it is neat that they have a friend with something no one else has.

To conclude, I must tell you, it is nice to know someone who not only has MSUD, but who understands what I am going through. This is one journey I will always remember.

Diana is a busy, active girl. She also served on a panel at the Symposium. She and Jessica Shaffer were the teens along with two mothers on the panel, Sandy Bulcher and Kay Larsen, who fielded questions from the audience.

SHARING - LETTER FROM NICKY GUTHRIE

Details

Written by Nicky

Published: 17 July 2009

I would like to say how particularly interesting I found reading "Brock's Story" in the Family History section of the last issue of the Newsletter. Our elder daughter, Laura, is only five, but school has highlighted certain areas of difficulty, and these areas were precisely pinpointed in Brock's story.

"Organizational skills are poor... His poor impulse control and vivid story telling makes it difficult for Brock to make lasting friendships.. He requires fine limits and constant guidance, enjoys testing those limits and resists guidance, but at the end of the day he's the first one to kiss you good night, give you a hug and express his love in front of anyone present!" All these things could apply to Laura! In fact, I found the similarities so interesting that I photocopied the page and gave a copy to Laura's teachers at school. I felt that it may help them to understand her.

On the whole, Laura has enjoyed her first year at school, but she has had a very difficult time during the last couple of weeks because a friend rejected her. This friend told Laura in no uncertain terms (as children do) that she no longer liked Laura. Laura was so upset that she lashed out, quite violently at times. She surprised and frightened herself with the violence of her own reaction, and this caused more stress, leading to slightly raised amino

acid levels. This led to more "inappropriate behavior" or perhaps I should say, "unacceptable behavior."

We had to more or less re-teach Laura how to interact with and react to the other children. She was beginning to take the most innocuous, negative response as a rejection and to lash out violently as a result of her upset. We had to explain how this is not the way to deal with the situation, and to discuss with her how she could have reacted more positively. We explained that this is the road to possible friendships, and the other way is not. She agrees with us, and wants to do what we suggest. Laura might have been severely punished for the way she was behaving. We are lucky to have such a sympathetic and supportive school.

Our thanks to Celeste for articulating the things she did so succinctly. If she has any more advice we would be more than grateful for it. If anybody has similar experiences, it would be great to hear about them, and to learn how they cope!

- With much love, Nicky