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PERSONALLY FROM THE BRUBACHERS

Details

Written by Joyce Brubacher

Published: 20 July 2009

With this issue I finish covering our last Symposium except for the talk given by Samantha Whitney from Australia. I am keeping her interesting view of living with MSUD for the fall/winter issue. In that issue I would like to feature the teens and adults with MSUD.

I would like to hear from all of you young people with MSUD. In this issue we announce Eric Rudd's graduation from college and several others have sent in their high school and college announcements over the years. However, there are many of you we haven't heard from. Why not write a short note - or a full-length article would be fine - about what you are doing now, how you manage your diet, how you deal with illnesses and maybe share how you feel about having MSUD or anything else you care to write. Those who have written before, send me an update.

Earlier this year I became acquainted with a 39-year old woman with MSUD. When we met Cindy Blau at Symposium '96, we assumed she was the oldest person living with MSUD in the U.S.A. and possibly in the world. Then Emily Talley contacted us, and she is almost two years older than Cindy. Emily has a mild form of MSUD and controls her levels through diet alone.

I have enjoyed getting to know Emily through e-mail. She has recently been accepted to graduate school at the University of Minnesota to pursue an MS degree in software engineering with applications to database design and networking. But I will let her tell more about herself in our fall/winter issue.

Shayla is finally rid of her headaches and is much more energetic than she had been for years. What made the difference? We finally found the key to lowering her leucine level. For many years her leucine level stayed within the 7 to 9 mg/dl (550 to 700 µmol/l) range. Trying to lower her already very restricted dietary protein only made her hungry, irritable and more tired. Except for periods of stress, all three levels seemed to stay very constant. After the last Symposium she was determined to try adding isoleucine and valine

supplements to her formula. Although she still tolerates about the same amount of protein, her leucine gradually dropped to the 3 to 4 mg/dl (250 to 300 µmol/l). She looks, thinks, feels and acts better and has finally lost some weight. We are all pleased with the results.

Shayla is now washing dishes part-time at a local restaurant. It is a demanding job during the tourist season. She made the mistake of thinking she was too busy to take time to eat on her first day alone on the job. She worked almost 10 hours and was dragging by the time she came home. She was tired and had a headache the next morning. Blood sent for testing the next morning revealed a level of 12 mg/dl. She also started her menstrual period that afternoon after blood was taken, so we are not sure if the elevation was solely from the long fast. She learned a lesson and now grabs a bite to eat along with her formula sometime during work hours.

I think the article on nutrition in this issue has some very important information. It is good to discuss the nutritional status of MSUD patients with your doctor and dietitian.

- Wayne, Joyce, and Shayla Brubacher

SYMPOSIUM '96 REVIEW

Details

Written by Joyce Brubacher

Published: 20 July 2009

(Continued from Vol., 14 No. 2)

The 1996 MSUD Symposium was held in Columbus, OH on June 20 through 22, 1996. This was likely the largest group of persons with MSUD to ever gather in one place. Symposium '96 was attended by 55 families who have children with MSUD.

The last issue of the Newsletter included highlights of the speeches presented the first day of our Symposium. This issue covers the talks given on the second day (Saturday). A mother and daughter from Australia and a mother from Chile shared their personal experiences. A professional and a sibling addressed sibling issues; the latest in diet and nutrition were covered by nutritionists and parents in a speech and a panel discussion; parents of older children with MSUD met for a time of sharing; and there was a talk on self esteem. During the Symposiums, parents have the opportunity to share their observations and concerns with other parents on an informal basis. This valuable part of the Symposium cannot be covered in the Newsletter.

NUTRITION PROBLEMS OF CHILDREN UNDERGOING THERAPY FOR MSUD

Details

Written by Joyce Brubacher

Published: 20 July 2009

Presenter: Phyllis Acosta, DrPH, RD

The following article summarizes the informative speech given by Dr. Acosta, Director of Metabolic Diseases at Ross Products Division in Columbus, Ohio. She reviewed the most recent nutritional information relating to the health of children with MSUD. This is important information for families to discuss with their nutritionists.

Nutritional problems in the early treatment of MSUD

Before there were medical foods (formulas) for MSUD, many children died. There was no way of treating them except to restrict protein. When protein is severely restricted, children stop growing, consequently reducing their need for amino acids, since they no longer need them to make protein.

Early nutritional problems with homemade and commercial medical foods:

- *Low blood sugar*- Compounds made by the patients with high keto acid levels interfered with making blood glucose.
- *Folic acid deficiency*- The fat soluble vitamins were first given separately. Parents sometimes forgot to give them, so they were added to the medical foods.
- Acidosis- This affects growth and bone mineralization. The first amino acids used in formula had hydrochloric acid attached to them, so they dissolved in liquid more readily. This caused acidosis and poor growth. (The sludge at the bottom of the bottle or cup of formula contains some of the amino acids and minerals. Be sure your child gets that sludge.)
- *Selenium deficiency*- It was not known to be an essential nutrient until reported in Germany in 1975.
- *Growth retardation*- It is a serious problem in MSUD and PKU. We are just starting to understand it, and it can be prevented.

The development of commercial medical foods

Between '61 and '78 clinics were mixing L-amino acids with vitamins, minerals, gelatin and some carbohydrates to make a medical food for MSUD. MSUD Diet Powder was the first commercial medical food marketed in '78. It was much lower in protein than what Dr. Acosta preferred; however, the pharmacist at the hospital where she was working refused to make the "horrible stuff" any longer when he learned a commercial medical food was available.

The first formulas had no quality assurance. Companies that make medical foods have to meet strict quality assurance. The nutrient composition must be maintained until the end of the shelf-life of that product. Quality assurance and costly ingredients, like the L-amino acids, add to the expense.

In the 80s, Scientific Hospital Supplies, a company in Europe, began marketing Analog, Maxamaid and Maxamum MSUD. In the 90s, Dr. Acosta had the pleasure of designing the products called Ketonex-1 and Ketonex-2 marketed by Ross Laboratories. Being a nutritionist and having worked with metabolic patients since the 50s, she wanted to design her own medical foods. She didn't like any of the products on the market at that time.

Deficiencies and growth problems
Reports from the 70s showed patients were not above the 25 percentile in weight or height. We are still seeing some problems with growth, some protein/energy malnutrition, and deficiencies in isoleucine, carnitine, and selenium.

Dr. Acosta showed charts of the protein and calorie intakes of children with MSUD compared with the Recommended Dietary Allowance (RDAs) and with a control group of children (average children) ages 1 to 11. Although the protein intake seemed adequate for children with MSUD according to the RDAs, it was much lower than the intake of the average child in the United States especially in the 4 to 11 age group.

A study reported in '90 involved 12 children with MSUD in the age range of 2.8 to 11 years. Their protein intake was 78% of RDAs and their calorie intake 86% of RDAs. They were shorter than normal and were selenium deficient. Selenium deficiency occurs anytime protein is restricted in the diet.

It was reported that three children with MSUD from England were significantly below normal in growth. There is a problem with the way we are feeding our children. If children are fed enough protein, they will grow normally. If we can get these children to grow normally, they will tolerate more BCAAs (branched chain amino acids). Growth in length is the best indicator of protein status.

Isoleucine deficiency makes children appear to have kwashiorkor, a severe protein-energy deficiency. Many infants with MSUD develop lesions on the buttocks, a sign of isoleucine deficiency. Unless blood is drawn every day when a clinician is trying to drop the BCAAs rapidly, it is hard to tell exactly when to start adding isoleucine. Adding isoleucine will eliminate the rash on the buttocks.

Carnitine is produced in the body but the nutrients must be provided in the diet. Studies of plasma-free carnitine levels during the pregnancy of a woman with MSUD revealed a need for larger than normal amounts of carnitine. This suggests children with MSUD may be losing carnitine because of some intermediate compounds they are making. It may be helpful to know what your child's plasma-free carnitine is.

Selenium, according to some very recent information, controls one of the enzymes in the body that changes a non-active form of thyroxine (a hormone produced by the thyroid gland) to an active form. A German report in the 70s indicated low levels of selenium in children with MSUD and PKU. In a study in Ireland, heart rhythms of PKU patients deficient

in selenium became abnormal and life-threatening. In a recent study, PKU patients were given selenium. This decreased their non-active levels of thyroxine and increased the active form.

If children are not getting enough selenium, they may not have enough of the active form of thyroxine. Thyroxine can affect bone mineralization, growth and IQ. Selenium is also important for the immune system. The soil is deficient in selenium in many areas of the United States. Foods grown in those areas do not provide enough selenium.

All the medical foods made in the United States have adequate selenium. MSUD 1 and MSUD 2 are made in Germany and do not have added selenium. So if your child is taking enough of a medical food made in the United States, he/she is getting an adequate amount of selenium. The companies have to put more than normal amounts of these products in the medical foods to get normal plasma concentrations.

Studies show, however, that children with MSUD ages 1 to 4 were getting 66% of RDAs of selenium but only 20% of what a control group of children was ingesting; ages 4 to 7 were getting 53% of RDAs and only 15% of what the control group was ingesting; and ages 7 to 11 were getting 17% of RDAs but only 7% of what the control group was ingesting. This suggests that children ages 4 to 11 were not getting enough medical foods.

What are some important factors that influence the IQ of a child with MSUD? A reference by Dr. Paige Kaplan (at Children's Hospital in Philadelphia) names three possible effects: how long the child suffered the insult of high branched chain keto acids before diagnosis, the long term metabolic control (how tightly the diet is controlled), and possibly the metabolic control at the time the test was done.

Recommendations for providing adequate nutrition

1. The protein in medical foods is an artificial form of protein. The amino acids are in their free form (like in meat already broken down by the digestive tract) and they are quickly absorbed. Because the amino acids are absorbed faster than the body can manufacture its own protein from them, the amino acid - after the amino group is removed - is used for energy purposes and not as a building block for protein.

   So we have to give the children more amino acids and feed them more frequently - four to eight times daily. It is not a good practice to give the medical food only in the morning and evening. Spread it out during the day and give some natural protein with the medical food. You wouldn't give your "normal" children all their food for the day at one time.
2. The three BCAAs are not in the same proportion as in natural protein (from foods). Leucine is higher than isoleucine and valine. Therefore, children with MSUD can become deficient in isoleucine and valine. By adding the pure isoleucine and valine to the medical food, you can enhance growth and prevent low levels of these two amino acids.
3. It is important that everything prescribed be ingested each day. Dietitians make parents very compulsive because it is important.

4. Certain things need to be monitored: nutrient intake, growth, plasma amino acids, albumin, and ferritin - an indication of iron status. If the medical food given does not contain selenium, a supplement needs to be given every day.

DIETARY PANEL

Details

Written by Joyce Brubacher

Published: 20 July 2009

Presenters: Phyllis Acosta, DrPH, RD, & Steve Yannicelli, MMSc, RD
MSUD Parents: Anne Fredericks, Tish Fuller & Glenda Groff
Following are thoughts and information shared by the panel in answer to questions from the audience.

The idea of giving the medical food throughout the day was emphasized. It is very rapidly oxidized if taken all at once and is used for energy instead of growth. If you are giving the medical food in one feeding, your child will not be growing as he would if you were giving it in three or four feedings. It is not complete unless natural protein (from foods) is given with it. One mother feeds her child 1 to 2 oz. of medical food every hour day and night when he is sick as a preventative measure to keep him out of the hospital. This may not work for all children with MSUD.

Cardiomyopathy has been associated with selenium deficiency. In an area of China that is severely selenium deficient, children die of cardiomyopathy.

Carnitine by mouth can cause diarrhea, but otherwise large amounts are harmless. Pharmaceutical grade is very expensive and not likely to be given in large amounts. The percentage of carnitine in the cheaper variety available in the health food store varies considerable and it is not wise to buy it.

Selenium is toxic in large amounts. It can be tested by measuring either plasma selenium concentrations or the concentration of an enzyme in the red blood cells that requires selenium to function.

There was a discussion on dental problems for MSUD. Here are some of the conclusions:

- When held in the mouth, formula (medical food) carries the same risk for dental caries (causing cavities) as milk.
- It is important to brush teeth after drinking formula.
- Some of the children actually have good teeth - flouride helps. Many have sensitive teeth for no known reason.
- Families handled wisdom teeth extractions differently. Some teens with MSUD had anesthesia; others were given locals. One was on glucose IV's for one

hour prior to anesthesia and extraction, during extractions, and one hour after surgery. She did very well. Some teens were put on sick day formula before extraction to prevent elevation of levels. No serious problems were reported.

Questioned about B vitamins in the formulas, parents were assured the Food and Drug Administration has an Infant Formula Act which specifies the range of levels of vitamins that have to be put in infant products. The MSUD formulas contain all the known required vitamins. (It is the amino acids and not the vitamins that make the formulas taste bad). Some parents may not be giving their children enough fat. Your child needs some cholesterol. One doctor advised his parents to switch their children from margarine to butter. Over restriction of fat causes growth retardation. It is important to have a balance of protein, fats and energy.

When there are symptoms of an impending illness, increase carbohydrates (calories), decrease natural protein and continue the formula even if you need to freeze it and give it as ice cubes. Try to keep calories and fluid intake high to prevent catabolism (burning muscle protein). The child may need more sodium when sick, because the diet powder is low in sodium. Adding sodium to the formula can cause serious problems, so check with your doctor.

SELF-ESTEEM

Details

Written by Joyce Brubacher

Published: 20 July 2009

Presenter: Emilio Amigo, PsyD
Dr. Amigo is a licensed psychologist who works primarily with children with ADHD or those involved with abuse or divorce. Life-sized outlines the youth had traced of each other on paper were displayed on the wall during Dr. Amigo's presentation. The families could take these home. These outlines were created earlier in the day in Dr. Amigo's workshop on self-esteem for the teens and young adults with MSUD.

Dr. Amigo began by explaining the life-size outlines the youth made in the self-esteem workshop he conducted. The outlines identified their personal boundaries. To help them understand self, they were asked to follow certain instructions. For example, they put the names of family members and others inside or outside the outline depending on how they felt about those persons. In various ways they illustrated their attitudes, limits, choices, talents, desires, values, etc. on the outlines, thus expressing how they felt about MSUD.

To illustrate the concept of esteem, they were to connect a price tag to the body outline and put in the amount they thought they were worth. First he explained to them that they were

unique and one of a kind which is often valued as priceless. Some of the youth couldn't identify with that. Others thought they were worth "BIG BUCKS."

Practical Tips for Developing Your Child's Self-esteem:

- Make one-minute connections with your child
- Write love notes to your child
- Be a model to children by taking an honest self-look
- Refrain from the use of negative communications
- Make a life celebrations book – each family member writing what they want to celebrate
- Give your child a "gift" every night
- Use the human touch – hugs, pat, reassurance
- Mutually tell stories together with children
- Share dreams you had and dreams for the future
- The paying attention game – see details, smell the roses
- Play music together – don't need to be talented
- Together visually plan the next day

Know the top stressors in your child's life; teach them coping strategies:

- Self-affirm, then affirm your child – don't play the blame or shame game.
- Help them set SMART goals – Sensible, Measurable, Attainable, Realistic, within a Time line.
- Help them manage time.
- Have them create a self-journal: use a special pen, use a theme.
- Do art; read and listen to music together.
- Instill positive memories; faithfully keep up photo albums.
- Foster healthy play. Maybe do something fun with food to balance the seriousness of food in their lives.
- Regularly interview each other. Keep learning about your children because they change.
- Empower not overpower – prepare them for life.

Teach them conflict-resolution skills:

- have a clear agenda
- communicate a belief in resolution – can be resolved
- one person talks at a time
- have empathy for the other person
- use "I" statements instead of "you" – sandwich statements (positive, negative, positive)

As parents, be REAL – model honesty, be willing to learn, be emotionally vulnerable, be willing to not know the answer, share your dreams and sorrows.

- Share the "top 5 things I want and need" with family members.
- Chronicle your child's life – keep things for them: scrapbooks, papers and drawings, note experiences and accomplishments.
- Have "no reason at all" days. Do something fun without a reason.
- Create your own holidays like "Son Day."
- Celebrate a "You Day" – family honors one child all day.
- Participate in family ministry and/or service.

Dr. Amigo answered questions from the parents. One question was: "What characteristics came out in your discussions with the children?" He answered by naming several: Some of the children viewed themselves as special because they have MSUD. In the workshop they

worked hard, took it seriously, laughed a lot and helped each other. They were concerned about who would see the outlines. They seemed to be aware of the normal developmental stuff for their ages. They had a good moral consciousness. Most of them drew their parents as persons who meant the most to them.

SIBLING ISSUES

Details

Written by Sara Kiel

Published: 20 July 2009

Presenter: Sara Kiel. Sara is the daughter of Carl & Sandy Kiel from Jenison, MI. She is 9, years old.

Hi, I'm Sara Kiel. I have a sister and 2 brothers. My sister Jenna, who is 5, and my brother Jesse, who is 3, have MSUD . . . and then there is my other brother, Adam, who is 11.

It was scary when I first found out Jenna had MSUD. I didn't understand what it was because I was only 4 at the time. As I grew up, I learned more about this disease and what Jenna could and could not eat. I also know that she has to drink her formula. In the last three years we have had Jesse around the house, which not only means another pain but another brother to drive you crazy.

Jenna has come to a stage where she started drinking her formula out of a cup, and this year she got a new formula. She gets stubborn and won't drink it. It is a pain to get her to drink and boring to sit there counting every little sip she takes. Now that Jenna goes to preschool, mornings just get a little bit busier trying to get her to drink. Being a sister to Jenna and Jesse means telling baby-sitters what they can and cannot eat, which formula goes to whom, and how to get Jenna to drink her formula. I have to tell my friends about MSUD when they ask.

When either Jenna or Jesse goes in the hospital it makes me feel worried, but I also feel happy for them because they are getting the treatment they need. It is fun to go to the hospital because there are neat pictures hanging on the wall. Near Christmas they have gingerbread houses. I also like to go to the playroom, cafeteria, supply room, and the mezzanine. The mezzanine has a stained glass sculpture that turns. It's cool!

Mealtime at our house is different than at most, because my Mom has to cook two meals. I like baking Lo Protein cookies and brownies with my Mom. At supper time Jenna and Jesse usually eat a potato, rice, or macaroni without the cheese and milk. And they have to put ketchup on EVERYTHING! Going out to eat means getting lots of fries and having Jenna beg for the pickle off everyone's hamburger.

Some experiences I have had include mistaking Jenna's formula for my cup of milk in the fridge. The flavor gave me a little shock there! I guess you get used to the taste after awhile.

To me, it is fun having Jenna and Jesse as a brother and sister because at Halloween I get to trade my smarties for chocolate candy. Another fun time was when Jenna and Jesse had their pictures taken for the Mead Johnson Special Kids Calendar. We got to meet photographers from California. On that same day I got to skip school and go to the Mead Johnson formula company and see their formula being made. They gave us a tour, and we all got a cool hat and other stuff.

In school Jenna is like anyone else except she brings her own snack every day. This year my class read to Jenna's preschool class, and I got teamed up with Jenna. I read books and we did art projects together sometimes. We made a book about her favorite foods. Jenna's book included potatoes, macaroni, and pickles, of course! Sometimes life gets a little bit crazy with formula and all this MSUD stuff, but actually having people in the family who have MSUD isn't that bad!

Sara was encouraged to honestly tell how she felt about her siblings with MSUD getting so much attention when they were sick. Sara's mother assured Sara she wouldn't be mad or upset at her for her answer. Sara admitted that it sometimes made her "MAD." She frankly answered a number of other personal questions from the audience. Thanks for bravely sharing with us, Sara. We parents wonder about the feelings of siblings, and it is wise to tell us, even though we may feel quite helpless to change the situation. Sara did a great job with her presentation. She and the next speaker, Vicki Delaski, were delightfully humorous.

SIBLINGS: IS THIS BEHAVIOR NORMAL?

Details

Written by Joyce Brubacher

Published: 20 July 2009

Presenter: Vicki M. Delaski, MS, LSW
Vicki is the mother of a son with autism and a daughter from whom she has learned so much about sibling issues. She supplied the following article in which she covers the key points of sibling issues. This is not a summary of the speech she gave at the Symposium, but it covers the same issues.

To understand issues that may arise between siblings and the brothers or sisters with disabilities, we must first look at sibling relationships in general. Our siblings know us better and longer than anyone else we will ever know. Our parents will know us about 40 to 60 years, and we won't share everything with them. As a matter of fact, if you're like me, there will be quite a few things they will never know, or you would still get a spanking.

Sibling relationships, on the other hand, can last up to 80 years. Our siblings are the people we experiment on with our new found talents (like lying, and the left jab or karate kick we saw on TV last night, OR our first batch of cookies, and the discovery of makeup). We laugh, we cry, we fight, we celebrate, we mourn, and we share our deepest thoughts, feelings, secrets and fears with our siblings because of the bond that is there from the beginning. They are our first social network, and we learn how to interact with others through our interaction with them.

Do we always get along with our siblings? No way! Do we always love them? Yes. Do we always know that we love them, or they love us; or are there times in our relationship that we are sure we hate them or they hate us? Do our siblings embarrass us or we them? The answer to all these questions is generally - yes! Siblings and our family are the foundation on which we build our self-concept and our people skills. They are the gauge by which we measure our successes and our failures.

Now let's talk about the relationship between siblings and their brothers or sisters with disabilities. The relationship and all the emotions are the same with one difference. The disability has a way of magnifying all emotions, especially guilt at feeling any negative emotion like embarrassment, anger, jealousy and resentment. The research states that about half the siblings feel that having a sibling with a disability was the worst thing to happen to them, and half feel it was the best. So, how, you might be asking yourself, do I know if there is a problem, and how do I solve it? Each child and situation is different, so there is no one way to tell if a problem exists.

Some identifiable warning signs are listed below.

- *Depression:* change in sleeping or eating habits, a sense of helplessness or hopelessness, continued irritability, has a difficult time concentrating or making decisions, may withdraw from social situations, doesn't seem to have any fun any more, and is negative about themselves or talks about hurting themselves.
- *Anxiety:* worries a lot, an increase in energy level without purpose, cries easily when frustrated, problems sleeping, worry about the health of family members, may have lots of headaches or stomachaches, and may be a perfectionist.

If you notice several of these that last two weeks or longer, you should discuss it with your pediatrician or a mental health professional.

Each child is an individual and will display signs of stress, confusion, embarrassment, jealousy, resentment, anger, loneliness, guilt and fear in a different way. Tom may become verbally and/or physically aggressive if someone teases him about his brother, but Ann may become introverted and shy. When there is a crisis at home, grades may go down and bad behavior may go up. Watch, listen, ask questions, talk to them, their teachers and their friends. Generally be involved and show an interest in their lives, so it will be easier for them to come to talk to you about their feelings and any issues or questions.

This brings up the question of how much to tell their teachers about the sibling with the disability. If there is a crisis, and the teacher is not aware of issues in the family, they cannot help you watch for symptoms that the student is having a problem. I can't stress enough the importance of keeping teachers informed and using them as resources.

Now, let's say you think there might be a problem. We all want to believe that we are the parent, and that our children will come to us if they have a problem or question. However, many children don' t know what to ask or how to ask it. They may feel that if they ask, it will make their parents feel bad, angry, disappointed or sad. The one thing they don' t want to do is add to the problem or situation.

The key to improving any relationship is communication. The best way to show that it is okay to talk about feelings is to model that behavior. Children may not always do as we say, but will generally do what they see us do. Let them know that negative feelings are not bad but are normal. Feelings are not good or bad - they are just feelings and we don' t have a lot of control over them. We can only control how we react to them.

Be open and honest. Spend special time with each child. Remember *fair* in the eyes of a child is much different than *fair* by adult standards. Evaluate the expectations you have of each child, and don' t forget to let them just be kids once in awhile.

Siblings need to talk about their feelings in a safe environment. That environment is viewed as safe using their eyes not ours. This means that it may not be at home or with friends, but with peers who are going through the same thing. Check with agencies in your area to see if there are any sibling support groups or workshops you and the child can visit. If not, look into starting one.

RESOURCES:
Sibling Information Network
Department of Educational Psychology
Box U-64 The University of Connecticut
Storrs, CT 06268 (203) 486-4034
Powell, T.H. & Gallagher, P.A. *Brothers and Sisters: A Special Part of Exceptional Families*. Baltimore: Paul H. Brookes Publishing Co., Inc., 1993

INTERNATIONAL ASPECTS OF MSUD, LORETO ILABACA, CHILE

Details

Written by Loreto Ilabaca

Published: 20 July 2009

First Presenter: Loreto Ilabaca, Chile
Loreto is Chilean and speaks Spanish. Although concerned about her English pronunciations, she did well when she gave her talk. She sent the following article for the Newsletter giving a little more detail of her experiences. It was written in Spanish

and translated. Several health care providers translate our Newsletter for their Spanish-speaking families. I can send copies of this article in Spanish on request. In her talk she mentioned there were six cases of classical MSUD and four intermittent (including her two children) in Chile. One with intermittent MSUD died last year. It is very hard for these families because of the lack of good hospitals and doctors. Special foods and formulas need to be imported and most families cannot afford the cost. Without the formula, the children eat only one meal a day and that is only vegetables. These children have many physical problems.

I am from Chile and the mother of two children with intermittent MSUD. Claudia is 14 and Christian is 12. The two children were born normal. When they were around two weeks old, they were diagnosed with severe Gastroesophageal Reflux. The doctors tried to correct this by using a postural treatment - sleeping with their cradle at a 40-degree incline. Because of the reflux, the children were always vomiting so much that I became accustomed to it, and it didn't bother me very much.

In December 1983 Claudia had her first vomiting crisis at 18 months. She was very sick. They did much testing, but the results didn't show anything strange. The doctors thought it was just a virus. When the vomiting didn't stop, they had to pump her stomach. When she started to recuperate, they hydrated her by giving her a spoonful of water every five minutes. This crisis lasted about four days.

The following year on the same day in December, Claudia had her second crisis and Christian, his first crisis. He was 10 months old. They gave new tests to the two children, but the results didn't show anything strange. The doctors told me they both had a virus. They hydrated them the same way as before.

The next December the children had vomiting crises again, repeating the experience of the previous year. Again we didn't find the cause of the crises.

Claudia didn't repeat these episodes until 1992 when she was dehydrated, and they had to give her serum. She has more enzyme activity than many MSUD, and so does not have as many problems. She learned she should be very careful with her diet when she is sick. Christian began to repeat these episodes. Some of them were caused by a viral sickness, during which he had to be hospitalized and given glucose serum, because he was totally dehydrated. This made the pediatrician suspicious of Ryes Syndrome and asked if Christian would undergo a series of tests. The results were negative. Christian repeated these crises for some months after he had these tests done. The pediatrician didn't understand what was happening, because he couldn't find the cause for these episodes. Many times they weren't accompanied by any contaminating illness and the results were always negative.

In February 1992 when Christian was eight years old, after various episodes in a short time, he had a very bad crisis with major head and abdominal pains. He was dehydrated and they had to give him serum for many days. The treatment didn't have any effect, and each day he got worse. They took a Cat Scan because of his bad headaches, and the results were normal.

The pediatrician suspected that he had a metabolic illness because of the characteristics of the crisis. He asked if he could give more specific tests. They sent Christian to a specialist

for metabolic diseases. She took another series of tests and diagnosed MSUD. They sent samples of Christian's blood to a Biochemical Genetics Laboratory at the University of Colorado Health Sciences Center in Denver. Twenty-one days later we received the results from the U.S.A. confirming the diagnosis - MSUD Variant, R10 E3 deficiency.

At this time, Christian's life and ours changed drastically. Christian, who consumed a great quantity of protein in his daily diet, was now put on a low protein diet. For eight years he had lived as a normal child and now had to learn to eat differently - things that he didn't like; and he didn't like vegetables. For Christian, this has been very difficult. He is a very brave child and makes great efforts to try to accept what has happened. However, many times he gets depressed, because he feels different and can't live the life that he lived before.

In Chile there is only one center where they can diagnose this illness. Every time we went there, we were very depressed and became very sad. When blood samples were sent to the U.S.A., many times the results didn't return for two months. I was thinking that this is not useful. One day I decided not to take him there anymore. I thought I could control his diet, and that everything was going to be OK. I would be eliminating a suffering experience for Christian. I spoke with the pediatrician and he told me it was my decision.

Our life changed a lot during that period. I think Christian felt happier not having to go to that place, and that comforted me. During those months he had various crises, but they were very small and didn't last long. With the help of the pediatrician they went away quickly. In November 1995, Christian became very sick, vomited a lot, had bad headaches and abdominal pains that led to dehydration. They had to give him serum at home.

This crisis lasted five days, but he began to recover. He was well for four days, and then had another crisis worse than the first. He was very sick, continuing to vomit a lot, was dehydrated again and had to be hospitalized. They gave him serum and we had to wait. I didn't know what to do when I saw him. They told me I had to wait. I felt very alone and I didn't understand and I didn't know if what we were doing was right. I didn't have contact with other mothers, and I felt terribly guilty for not having been more in control.

They had to take more tests and again the specialist told me that Christian had high levels of leucine, isoleucine and valine. They didn't want him to leave the hospital because the diet that had worked before wouldn't work anymore; he was growing and his metabolism was changing. Fortunately, he recovered. When we left the clinic, he began a new, stricter diet than before. He began to take Ketonex-2. He hated it, and we fought a lot to get him to take it. They also gave him carnitine and thiamine.

I was feeling very badly one day when speaking with the pediatrician. Then he told me that he had found the name of this association on a computer program referring to this strange disease. He put me in contact, because he thought it was the only place where I could find answers to my questions - with parents who felt as I did. I didn't doubt him and I called Peter Shaffer. I told him my history and he told me that he would send me information.

In February of this year I received my first Newsletter. I felt so happy reading the experiences of other parents who felt and lived like me. I wrote immediately about attending the Symposium. In May, I spoke with Sandy Bulcher, and she asked me if I would tell my experiences. I accepted immediately. I was a little frightened because of my English, and I

worried that no one could understand me, but I wanted you all to know my children and our history.

I also wanted to tell you that it was a marvelous experience, to be able to share with you my fears, sorrows, doubts, and to see the pretty children. For the first time in these difficult four years, I could speak with you and you would understand me, and not look at me as if I were from another planet.

I want to thank Dave and Sandy Bulcher for having me at their home and making me feel as if it were my home; also Wayne and Joyce Brubacher, Phoebe Saunders, Brenda Wenger, Tanya and everyone else who helped me. Unfortunately I can't remember everyone's name. Thanks to all of you for giving me the opportunity to meet you. I feel that in two days I learned more than I was able to learn in four years.

I am a very overprotective mother. When I left Chile I was a little worried because I didn't know what would happen in my absence. When I returned, I was very happy and felt at peace with myself. I could feel assured that everything was all right, and I could do things that only mothers can do. My children understood this. I want to say thank you to Joseph Balinsky, because very recently he told me that the best gift I could give my children was to teach them to be independent.

NEWS & NOTES - A COLLEGE GRADUATE

Details

Written by Joyce Brubacher

Published: 20 July 2009

Nikolai Rudd (classic MSUD) graduated with a BA degree on May 10, 1997 from Guilford College in Greensboro, NC. Nikolai, who lives in North Adams, Massachusetts with his parents, Barbara and Eric Rudd, majored in Theater Studies.

Nikolai was especially busy during his last year, serving as the film coordinator for the Student Union and president of the Guilford Revelers, a student drama organization. Through this group, Nikolai spearheaded the production and was director of the play, "A Few Good Men." (The movie by that name starred Tom Cruise.)

While attending college, Nikolai was able to maintain his diet satisfactorily. His parents arranged a close cooperation between Dr. Jerry Joines, a Greensboro doctor, Dr. Stephen Kahler, a specialist at Duke University Hospital (one hour away), and Nikolai's regular doctor, Dr. Vivian Shih, at Massachusetts General Hospital. Although Nikolai had to go to

the local hospital a few times for overnight IV treatments during his first three years of college - done as a preventive measure when he was coming down with a cold - no further medical attention was needed.

Nikolai was able to fully participate in college activities. The college agreed to a greatly reduced board fee because of the limited amount of food Nikolai was able to eat in the college cafeteria. The MSUD formula was sent directly to Nikolai who prepared it in his room. He is presently back in North Adams where he is pursuing employment in his field.

Congratulations, Nikolai, from the MSUD Family Support Group. We wish you well.

NEWS & NOTES - REPORT ON BEHAVIORAL STUDY

Details

Written by Joyce Brubacher

Published: 20 July 2009

Chip Kobe, PhD, director of the Columbus Center for Behavior and Learning, Gahanna, Ohio, spoke on ADHD IN SPECIAL NEEDS CHILDREN at Symposium '96 (see MSUD Newsletter, Vol. 14, No. 2, p. 6). While preparing for the talk, he found little published on the subject of behavioral aspects of MSUD, so he decided to conduct his own study.

Last fall Dr. Kobe sent questionnaires to all the families on our mailing list who have children with MSUD, age 2 to 18. Of the 160 distributed, 85 were completed and returned. He was pleased with this response and is in the process of tabulating and reviewing them. So far the questionnaires have revealed a high frequency of attentional and social problems.

We parents have noticed common behavior patterns in many of the children with MSUD. We have discussed these behaviors informally at the Symposiums and shared through the Newsletter. Not all the behaviors create problems, but some can interfere with routine living and academics. Now the professionals are taking note.

Dr. Kobe will make the results of his study available to Dr. Dean Danner, a professor in the Dept. of Genetics at Emory University, Atlanta, Georgia. Dr. Danner spoke on gene therapy at Symposium '96. For years he has researched the genetics of MSUD and is now interested in studying the prevalence and origin of behavioral problems in the MSUD population. Are the behaviors a result of the diet treatment, the enzyme defect, or are they related to episodes of ketoacidosis that occur during illnesses?

Wouldn't it be a tremendous relief to know the problem behavior some of us parents have tried so earnestly to deal with is not just poor parenting? Identifying the origin is the first step in finding a way to prevent or improve this behavior in order to help these children live better lives.

A sincere thanks to all of you who took time to complete the questionnaires and cooperate in this study. As Dr. Danner said at the Symposium: the families make research possible. To fund and conduct a scientific study, Dr. Danner will need more help from the families as this behavioral study gets under-way. We have confidence you families are interested in this study and will continue to cooperate. We wish the researchers well.

NEWS & NOTES - SUE ANN HAS A SECOND CHILD

Details

Written by Joyce Brubacher

Published: 20 July 2009

In our Dec. '90 issue of the Newsletter we announced the birth of Amamda McKnight, daughter of Lon & Sue Ann McKnight. Amanda's birth was a special medical event. Her mother, Sue Ann, has a type of classic MSUD. She was the first mother with MSUD to be reported in medical literature. Amanda was a full-term, healthy baby who attended kindergarten this past school term. She is a perfectly normal little girl. Amanda now has a brother, Blake.

Blake came early. He was 29 weeks premature and weighed only 1 lb. 4, oz. Born March 6, he came home from the hospital on May 29 weighing 3 lb. 15.8 oz. Sue Ann intends to write about her experience for the next issue of the Newsletter.

Congratulations Lon and Sue Ann. We are all eager to hear your experiences and how Blake is doing. May the Lord bless you all with health and happiness.

NEWS & NOTES - FROM SICKNESS TO HEALTH

Details

Written by Joyce Brubacher

Published: 20 July 2009

The Norman Burkholder family, from near Kutztown, Pennsylvania, recently had a very stressful experience. It all began when the Burkholders, in consultation with a local homeopathic doctor, gave their 8 year old daughter, Kathryn, large doses of vitamin A in a sincere effort to boost her immune system. Her formula also contained vitamin A. Norman and Mabel didn't realize that giving her mega-doses of the vitamin for several years would ultimately destroy Kathryn's liver.

When the cause of liver failure was determined, the vitamin was stopped. They hoped Kathryn's liver would heal. . . . It didn't. On the way to the Clinic for Special Children at Strasburg, PA on Jan. 2, 1997, Kathryn brought up blood - a sign of end-stage liver failure. She was admitted to the Lancaster General Hospital under the care of Dr. Holmes Morton. The family had to make a decision - she would die without a liver transplant. They decided to try the transplant.

Metabolic control was maintained at the Lancaster hospital until she was ready for the transplant. It took place at the Children's Hospital in Philadelphia on Feb. 12. Kathryn was the first person with MSUD in America and the second MSUD in the world to have a liver transplant. It was successful according to Dr. Paige Kaplan, a geneticist at Children's Hospital.

Kathryn came home from the hospital on March 21. By the middle of April she had grown an inch and gained 5 pounds. She is growing strong and robust. Why wouldn't she? SHE IS NOW EATING MEAT AND CHEESE SANDWICHES AND TOLERATING A NORMAL DIET. The new liver evidently provides some enzyme activity. Whether the cure is complete remains to be seen. After a 12 hour fast, Kathryn's blood test showed a small amount of alloisoleucine (only seen in MSUD). However, she seems to be tolerating a normal diet and is the envy of her younger sister with MSUD, Ella Mae, 4.

Before we even think of a transplant as a solution to MSUD, there is much to consider. Most patients with MSUD have healthy livers. The operation is risky and astronomical in cost. Transplant patients need to be on very costly anti-rejection medication the rest of their lives. Kathryn's medications are more expensive than the MSUD formula. Relieved of one set of problems, she faces others.

Doctors have learned much from Kathryn's experience. The liver may even prove to be a target organ for gene transplant, although there are still many difficulties to overcome. We parents can also learn from this. A nutritionist is a critical part in the care of MSUD and needs to know exactly what individuals with MSUD are consuming in their daily diet.

The Burkholder family does not carry medical insurance for religious reasons. Neighbors and friends held a charity auction and raised $27,635 toward paying the $500,000 bill. Local churches also gave generously, but a huge bill remains. If anyone wishes to help, contact

the Burkholders or send donations to the Kathryn Burkholder Liver Transplant Fund, 224 Dotterer Rd., Lenhartsville, PA 19534-9584.

NEWS & NOTES - IN SYMPATHY

Details

Written by Joyce Brubacher

Published: 20 July 2009

We learned of the sudden death of Leroy Martin, the son of Lester and Anna Mae Martin, Barnett, Missouri. He was one of their six children and the only one with MSUD. Leroy had been doing really well lately. He was almost 8 years old and helped chore on the family dairy farm. He rode his bike 1, miles to school each day attending first grade.

On Feb. 28 Leroy vomited all day and all night. The next several days he seemed better with only occasional vomiting. He continued to drink his formula and felt good enough to do school lessons again by the end of the week. Then on Mon. morning, April 7, the vomiting started again. He dressed himself that morning, but at noon they took him to the hospital. When they got there, he was stiff and dehydrated but alert. He died quite unexpectedly early the next morning, April 8.

We extend our sincere sympathy to the grieving family. Life changes so dramatically with the loss of a special child. Let us remember them with cards and prayers.

FAMILY HISTORY - ZACHARY PINSKEY

Details

Written by Denise Pinskey

Published: 20 July 2009

Zachary was born on Sept. 6, 1995 to Ron and Denise Pinskey. He was a beautiful boy who nursed great before we left the labor delivery room. He had a sister, 19 month old Alexandria. We could not have been happier - two beautiful children who appeared normal.

Unfortunately, a lot has changed since that day. Zachary stopped nursing on the fifth day of life. By his seventh day, a breast feeding specialist said Zachary kept his tongue on the roof

of his mouth and sucked on his lip; that was why he could not nurse. She gave me some instructions and we were on our way.

That same day I received a telephone call from the doctor who delivered Zachary. She explained that he tested positive for maple syrup urine disease, which can cause brain damage and death if not treated quickly. Needless to say, everything was a mess. I couldn't reach my husband because he was at a training class for work and I didn't know the number. I had to drive Zachary to the local hospital for a blood test. Then at the doctor's office, I had to wait for an hour. Our doctor was unsure if his diaper smelled sweet. She had never seen anyone with this disease, so she wanted me to go to the University of Michigan. It was almost two hours away and I had never been there. Fortunately, our luck changed, and my sister was able to drive us to the hospital.

When we got to the hospital, Dr. Richard Allen smelled Zachary's diaper and felt very positive that he had MSUD. He explained that the disease is treatable. During this time Zachary had his eyes closed the entire day and didn't open them up for a day or two. Unfortunately, Zachary's levels took their time coming down, and he had lost his suck reflex. He was hospitalized for two weeks and went home on a feeding tube. The feeding tube was a horrible contraption. Zachary's condition did improve. But within a month, he got a cold, which turned into pneumonia caused by aspirating due to the cold and the feeding tube. Fortunately, his levels did not rise with this illness. After a few days in the hospital he began sucking from a bottle full time. YEAH! It has been a year since he was hospitalized with the rota virus, which was as hard on him and us as when he was diagnosed at the hospital.

Zachary has steadily improved. He sat up at 9 months and crawled at 10 months (combat crawl). He walked around furniture for 4 or 5 months before he finally let go. He walks on his own more and more, but he really likes to walk on his knees. Zachary likes to feel very safe, so that's why he walks on his knees and eats very basic things. He is very social, loves people and smiles a lot. He is currently receiving physical, occupational, educational, and speech therapy through our county which has helped a lot. He is developmentally behind 4 to 6 months, which we hope he will overcome with good health.

Zachary has tested every belief we ever had, but he has also rewarded us with all his smiles, hugs and kisses. Right now he is very active and can't wait for summer to come, because he loves it outside and loves to swim. We once again feel that we are very lucky in life.