Liver Transplantation in MSUD: Protocol and Clinical Experience Including Domino Transplantation

George Mazariegos, MD
Children’s Hospital of Pittsburgh

Dr. Mazariegos treated attendees to an exciting update on liver transplantation as a treatment option for MSUD.

Medical therapy and transplant both carry risk. For the individual with MSUD, metabolic decompensation is always a possibility. As Dr. Mazariegos noted, the highly restrictive nature of the diet affects quality of life, and the cost of formula and low protein foods can be prohibitive for many. Transplant not only carries risks for potential complications rejection and the need for immunosuppression. The extent to which these latter risks can be minimized is a critical factor in the decision whether or not to transplant.

Liver transplant is a treatment choice for a number of disease states in addition to metabolic disease, including malignancy, alcoholism, biliary atresia, and hepatitis. Data (Mazariegos, M.D. cont. on page 4)

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The information contained herein does not necessarily represent the opinions of the MSUD Board, Medical or Nutritional Advisors, or all of our members. Before applying any of the information contained in this newsletter, you must consult a MSUD specialist.

Curing MSUD: Update on the Mouse Project

Gregg Homanics, PhD
University of Pittsburgh

Gene therapy offers hope of a cure for MSUD. To study the possibilities of this approach it is necessary to have an animal model on which to experiment. Dr. Homanics, Dr. Paul and their colleagues have created such a model with mice genetically engineered to mimic the pathology of the classic and intermediate forms of MSUD.

Prior to the development of the MSUD mouse, the only MSUD animal model was a cow. The mouse model offers numerous advantages over the cow including an amino acid metabolism which is similar to that of humans. Mice are a much more practical model, as they are inexpensive to raise and reproduction is rapid.

Dr. Homanic’s group first created a classic model of MSUD by inactivating the gene responsible for the E2 subunit of the branched-chain keto acid dehydrogenase enzyme complex using gene targeting and embryonic stem cell technologies. Embryonic stem cells were isolated from blastocysts and grown in culture. These cells were injected back into the embryo and implanted into a foster mother.

This process took several years. The resultant animals had (Homanics, Ph.D. cont. on page 4)
MSUD Symposium 2006 was held in Dublin, Ohio at the Embassy Suites on June 15-17th.

Approximately 250 people attended the three day event. MSUD families from 20 of the 50 United States were present. In addition, MSUD families and professionals from Canada, Brazil, Argentina, India, Honduras, Philippines, Costa Rica, and South Korea attended the conference. Seventy individuals with MSUD ranging in age from infant to 47 years old were present.

A reception was held Thursday evening in the beautiful atrium of the Embassy Suites. During that time, MSUD families enjoyed renewing old friendships and making new friends.

Friday was a full day with many topics and speakers, including:

- Research Update on MSUD Mouse Models          Gregg Homanics, PhD
- Current Status of Transplantation for MSUD     George Mazariegos, MD
- Improving Self-Esteem                          Darlene Honigford, LSW
- MSUD-Teen/Young Adult                         Jordan Bulcher, Libby Stone, Mark Silva, Galen Carrington, Alana Moceri

Breakouts (participants attended 2 of the 4 sessions, depending on their interests)

- MSUD Basics                                    Dennis Bartholomew, MD
- Sharing Daily Challenges                       Elaina Jurecki, RD
- Liver Transplantation, Parent Perspective      Denise Pinskey
- It’s So Easy-Low Pro Cooking                   Malathy Ramanujam
- MSUD Group Photo

Saturday was both informative and fun with a half-day of speakers and half-day picnic:

- New Developments in the Treatment             Holmes Morton, MD
- of Maple Syrup Disease                        Elaina Morton, MD
- Nutritional Management of MSUD                Elaina Jurecki, RD
- MSUD Board Update                             Elaina Jurecki, RD
- Closing Video
- Family Gathering at a nearby park for all symposium attendees. All present enjoyed carnival games, large climbing toy, volleyball, badminton, juggler and, of course, lots of protein-free candy.

Special thanks to United Services Foundation for their generous donation, which allowed many MSUD families to attend the symposium.

Plan to attend the next symposium in the Summer of 2008. Don’t miss the opportunity to learn from professionals and MSUD parents and feel the warmth and companionship of our special group!

**Sandy Bulcher**, Director, MSUD Family Support Group, Coordinator of MSUD Symposium
SYMPOSIUM SCRAPBOOK

JUNE 2006

www.msud-support.org
on survival outcome based on diagnosis, though, have shown that survival rates are highest for those with metabolic disease.

Thus far, 19 MSUD patients, ages 1.7 to 32 years, have undergone transplant. The first was a girl exhibiting vitamin A toxicity. After her transplant, her amino acid levels normalized without dietary restrictions. Subsequently, an additional 18 MSUD Patients (classic variant) were transplanted under elective protocol as described between 5/30/2004-5/31/2006.

While there have been complications post-transplant, all transplantees have survived and have normal liver function. A number of patients have experienced liver rejection, steroid sensitivity, and viral infections, however all have done well with medical and surgical treatment. Patients have experienced normal branched chain amino acid levels immediately post transplant, with maintenance of metabolic stability under conditions of unrestricted protein intake as well as inter-current illnesses. In addition to the obvious medical benefits, subjective improvements have been noted in attention span, speech, behavior and motor skills.

Historically, the heavy doses of immunosuppressive drugs required long term has had a significant impact on quality of life post-transplant. Cyclosporin, for example, causes excessive hair growth, weight gain, high blood pressure and bone disease. The ability to taper and, ideally, to withdraw immunosuppressive medications while continuing to prevent rejection is the ultimate goal. Tacrolimus, a newer class of drug, is moving treatment in that direction.

Variability in immune function allows some patients to be more successful than others in reducing their dependency on immunosuppressive drugs. Currently 28% of selected long term liver transplantees have been able to remain drug-free. It is hoped that an evaluation of the characteristics of these patients may allow others to achieve drug-free status.

The group hopes to develop an MSUD transplant registry to allow tracking of all who undergo this procedure. Sequential neuro-developmental evaluations are also planned to elucidate improvements which may be occurring in this area. The ability to transplant the MSUD patient’s liver into a non-MSUD patient (domino transplant) without transferring MSUD will benefit others as well.

Dr. Mazariegos suggests that individuals with MSUD and their parents consider the following potential indications for transplant:

- Lability of the disease despite optimal medical control and the development of metabolic crisis
- Neuro-developmental sequelae:
  - Attention deficits, hyperactivity in children
  - generalized anxiety and panic attacks, depression, delay in adults
  - Potential for cerebral edema
- Dependency on medical enteral formulation
- Intensiveness of medical therapy compared to post transplant therapy

Dr. Mazariegos acknowledges the assistance of his colleagues:


Children’s Hospital of Pittsburgh, Clinic for Special Children, Strasburg, PA
University of Pittsburgh Medical Center and the Thomas E. Starzl Transplantation Institute

Dr. Homanics discussed the value of stem cell therapy. He explained that, unlike viral vectors which address a specific enzyme defect, all subunits are present in stem cells. The result is that individuals with MSUD will be helped with this technology regardless of the specific nature of their defect.

Preliminary results using gene therapy with a single injection into the peritoneal cavity (belly) of the mice have been exciting, showing an increase in liver enzyme activity from about 9% to 30. The group plans further attempts using multiple injections, intravenous administration, and direct administration of the enzyme into the liver. Once treatment modalities are developed using the intermediate model, the group hopes to apply these modalities to the classic model.

The team is grateful for the support it receives from the NIH, Scott Foster Metabolic Research Fund, and the MSUD Family Support Group.
Effects Of MSUD On Learning And Behavior

Hilary Feldman, PhD
Child Development Unit
Children’s Hospital of Pittsburgh

It is clear that early diagnosis and treatment of MSUD is critical in order to prevent permanent physical and neurological deficits. In their review of the developmental outcome of 22 patients with MSUD, Hilliges, Awiszus, and Wendel (1993), postulated that cognitive outcome is associated with exposure to excessively elevated branched-chain amino and oxoacid levels during the postnatal period. They found that children treated before 5 days of age have a higher likelihood of having a normal IQ.

Nord, van Doorninck, and Greene (1991) also found that children who were diagnosed at a younger age had a milder neonatal course. Children with more moderate or severe complications had lower IQs than those who were asymptomatic or only suffered mild complications. Those children with milder complications also required fewer special education services when they became school-aged.

Mazur, Field, Berlin, Berry, Heidenreich, Yudkoff, and Segal (1991) found that children who had IQs below average had been diagnosed later than those with normal IQs (mean of 3.5 days for normal IQs and mean of 10 days for below normal IQ). Children who had been treated before they had severe symptoms had higher IQ scores than their siblings with MSUD who were not treated until they became symptomatic. The most important influences on IQ were age at the time of diagnosis and long-term metabolic control, although it was felt that control at the time of diagnosis may also have had some effect.

Morton, Strauss, Robinson, Puffenberger, and Kelley (2002), reporting on their experience managing a group of patients over 11 years, note that when classical MSUD is well managed in the neonatal period, individuals can have normal growth and development and relatively few hospitalizations. Unfortunately, common infections and injuries at any age can result in medical complications which can lead to a decline in neurological functioning.

As they grow older, learning and behavioral problems have been identified in children suffering from this disease. While not well studied, Nyhan, Wulfeck, Tallal, and Marsden (1989) found deficits with cognitive and language functioning among children with MSUD. Clearly there is variability, with some children exhibiting significant deficits while others fall within the average range.

Hoffmann, Helbling, Schadewaldt, and Wendel (2006) looked at the effect of long-term metabolic control on cognitive outcomes of 24 children with MSUD during their first 6 years of life. They focused on yearly medians of mean plasma leucine levels and identified three clusters of children: those with low, intermediate, and high levels. Those in the cluster with the lowest leucine levels had IQs that were statistically higher and close to the average range (median IQ = 102), while children with intermediate leucine levels had IQs which fell below average (median IQ = 76) and those with high leucine levels had IQs which were most significantly below normal (median IQ=57). They concluded that children can have normal cognitive functioning if long-term leucine levels are close to normal and if they have not had neonatal encephalopathy.

Cognitive and behavioral outcomes are assessed as part of the evaluation for individuals with MSUD being considered as transplant candidates at Children’s Hospital of Pittsburgh. Those who are accepted are evaluated again post-transplant. To date, 17 individuals have had psychological testing as part of the pre-transplant evaluation and five individuals have been evaluated post-transplant. We have looked at a variety of measures including cognitive functioning, receptive vocabulary, academic skills, verbal and visual memory skills, visual motor skills, fine motor speed, executive functioning skills, adaptive functioning skills, and parental report of behavior. Our findings support the literature indicating that there is significant variability in skills among individuals with MSUD.

Individuals evaluated as part of the pre-transplant assessment ranged in age from 17 months and 31 years. Individuals seen after transplant ranged in age from 3 and 27 years. Of the individuals who were evaluated as part of the pre-transplant protocol, IQs ranged from mental retardation to average. Of these individuals, 47% obtained scores within the average range, 29% obtained scores below the average range, and 24% obtained scores well below the average range. On testing of receptive vocabulary, scores ranged from well below average to above average, with 7% scoring above the average range, 64% of individuals scoring within the average range, 21% below the average range, and 7% of individuals scoring well below average.

On assessment of verbal and visual memory skills, individuals obtained scores ranging from well below average to above average. On a visual-motor task and testing of fine motor speed, scores ranged from well below average to above average. Assessment of academic and early learning skills indicated that 60% of individuals obtained...
scores within the average to the above average range and 40% of individuals obtained scores below the average range.

Parents also were administered an adaptive functioning measure which measures the individual’s ability to function in the daily world (based on what is expected for the individual’s age). Individuals are assessed on performance within the areas of Communication, Daily Living Skills (e.g. dressing, bathing, and other activities of daily living), Socialization, and Motor Skills. In the current sample, 50% of individuals obtained scores within the average range, 13% obtained scores below the average range, and 37% of individuals obtained scores well below the average range. Behaviorally, of 12 individuals, ranging in age from 6 to 31, over 80% were described as having or having had significant behavioral or attentional difficulties.

There have been five individuals who have been evaluated post-transplant with an age range of 3 to 27 years. Due to the wide age range and the diversity of individuals in the group, it is difficult to compare individuals. However, all parents and patients who were old enough to assess their experience, reported satisfaction with the transplant decision. One mother stated that it felt like a “burden had been lifted.” Another young adult reported that “I feel like a whole new man.” All individuals and/or parents reported satisfaction about being able to eat a variety of foods and not having dietary restrictions. All individuals and/or parents reported feeling less stress regarding ongoing concerns about the possible negative consequences of illnesses.

Although the sample is very small, parents of preschool-aged children who obtained transplants report being extremely satisfied with their child’s developmental progress post-transplant. Parents report that their children seem to be “brighter” and more alert. One preschool-aged child was functioning at or above the average range on most measures, whereas prior to the transplant the child displayed developmental delays. In older individuals, while there may be permanent deficits based on the child’s medical complications prior to the transplant, a trend toward better behavioral functioning has been observed.

Individuals with MSUD are a diverse group. Of our current, limited sample, almost 50% of individuals with MSUD have average intellectual functioning and almost 70% of individuals have average receptive vocabulary skills. On testing of academic skills, 60% of individuals obtained scores within the average to the above average range and 40% of individuals obtained scores below the average range. On an adaptive functioning measure, which assesses daily functioning based on parental report, almost 50% of individuals fell within the average range, although 37% of individuals were reported to be functioning well below the average range. In addition, a significant number of individuals (greater than 80%) are reported to have behavioral difficulties.

It is clear that continued study and research is needed to further understand the effects of MSUD on cognition and behavior and to identify the role that transplantation can play in minimizing further medical complications as well as increasing the quality of life for individuals and their families. One of the challenges is to continue to support families and to help families encourage age appropriate independent and adaptive functioning of individuals with MSUD.

REFERENCES


Nutritional Management of MSUD
Elaina Jurecki, MS, RD

The goal of diet therapy is to maintain branched chain amino acids (BCAA), particularly leucine, within normal limits as studies have shown a correlation between cognitive decline and elevated blood leucine levels. An aggressive treatment protocol during illnesses is also required for a positive outcome.

To briefly review of the metabolic pathways involved in MSUD, protein from food is digested and broken down into its amino acid components. The primary job of these amino acids is to make necessary tissues for the body such as muscle and heart. Amino acids may also be converted into blood sugars and ketone bodies. The BCAA are essential, meaning they cannot be made by the body and must be consumed from food. Normally, if more protein is eaten than is needed to serve the body’s needs, they will be broken down to make energy. In MSUD, however, the enzyme required is either present in inadequate amounts or is not functioning properly. As a result, leucine and its ketoacid accumulate to toxic levels.

The enzymes involved in the metabolism of the BCAA are large complexes containing 6 different components, coded by 6 different genes. The Carboxylase E1 unit needs the vitamin Thiamin as a co-factor with an alpha and beta subunit to make it function properly. Other components include Transacylase E2, Dehydrogenase E3, and kinase which is the activator of the complex. Phosphorylase inactivates the complex. A genetic disorder can involve a dysfunction in any or multiple parts of this enzyme.

The types of MSUD are related to the portion of the enzyme complex affected and to the degree of enzyme activity. The classical form of MSUD typically only has about 2% activity. Individuals with this form usually present in the newborn period with symptoms. These individuals require the most intensive form of treatment including special medical foods and formula with a very strict protein intake. Intermediate and intermittent forms of MSUD will have more enzyme activity and present later in life, usually from 5 months to 2 years. These disorders are not as common as the classical form. Because these conditions are milder, protein restriction does not have to be as severe, and frequently these individuals will not need a special metabolic formula. They may have to supplement their intake with special low protein foods to ensure adequate calorie intakes. Those with the thiamin responsive form only need to receive large doses of this B vitamin for management of their disease. A rare form of the disorder affects the E3 dehydrogenase complex and may present in the newborn period with lactic acidosis. Because of the many forms of MSUD, there is a great degree in variability in diet management. It is important to keep this in mind when reading the newsletter and other materials regarding diet and treatment of individuals with MSUD.

The effectiveness of treatment is monitored by nutritional status, metabolic control, and compliance. Metabolic control can be monitored at home by checking the urine for ketones with DNPH and tracking dietary protein intake. Nutritional status is monitored by monitoring growth, laboratory data, and by physical exam. It is quite important to closely follow the child’s nutritional status as there have been reports of malnutrition in patients with inborn errors of metabolism (Acosta P. SIMD / Molec Genet and Metab 81, 2004).

The goal of dietary treatment is to maintain blood levels of BCAA within the recommended limits while ensuring adequate calories and protein and avoiding nutritional deficiencies. This will promote optimal growth and development which will ensure quality of life. To accomplish this the diet must be restricted in the BCAA, specifically leucine, while delivering enough protein and calories. This requires the use of special medical formulas with supplementation from isoleucine and valine. Metabolic formulas include all of the essential amino acids needed by the body without the BCAA. These formulas will provide up to 90% of the MSUD child/adult’s protein intake. The variety of metabolic formulas available has significantly increased over the years. Several companies now make formulas for MSUD, including Ross, Mead Johnson, Applied Nutrition, Vitaflow, Milupa, and Nutricia. Infant formulas include Analog, Ketonex1, BCAD1 and are nutritionally complete. Formulas for older children, teens and adults include Acerflex, Maxamaid MSUD, BCAD2, Ketonex2. These formulas are more concentrated in protein and not as dense in calories. Applied Nutrition makes Complex MSUD and Complex MSUD bars which are higher in protein. Even more concentrated formulas such as Maxamum MSUD, Milupa MSUD2, and Vitafo’s products, MSUD Express and Gel are now available for use. The very first formula used to treat this condition, MSUD Diet Powder from Mead Johnson, has been taken off the market.

The protein in the formula is best used by the body when taken multiple times a day. A study looking at frequency of administration of metabolic formula showed that for the...
most optimal protein synthesis, the body requires the presence of amino acids at intervals throughout the day in combination with fats and carbohydrates. Amino acids cannot be stored and any surplus will be broken down to form energy. To become a source of energy, the nitrogen that is in amino acids is removed and excreted in the urine. A study of 10 young people, 12 to 26 years of age, with phenylketonuria, a more common metabolic disease involving similar dietary management, found that more nitrogen was wasted in their urine when formula was only ingested once a day versus multiple times a day. Consuming formula less frequently results in poorer use of amino acids for protein synthesis.

In addition to metabolic formulas, the use of protein-free formulas may be necessary to ensure an adequate caloric intake. There are several products to select from including Profree, PFD1, and PFD2 which are all vitamin/mineral fortified, and Dietary Specialties Calorie Supplement, Polycose, and Duocal which provide calories without other nutrients. Special low protein foods provide another source of caloric supplementation. There has been a significant increase in the number of companies that make these foods, and there are so many great recipes and cookbooks available to help improve the variety and quality of the diet.

Since protein and specifically the amino acid, leucine, must be limited, it is important to keep track of the exact amount provided from foods by using a food list. A number of sources are available, including an MSUD Food list developed by Emory University, Bowes and Church, and the USDA nutrient data base available at www.nal.usda.gov/fnic/foodcomp. A computer listing called MyDiet.com developed by Nutricia is also available. The Ross Metabolic Protocols provide guidelines for amount of protein, calories, BCAA, and fluids required per age group, but these guidelines must be tailored to meet the individual patient’s needs.

Estimates of the leucine content of foods can be made by using food labels to determine the amount of protein per serving and calculating the percent of leucine in a gram of protein. This varies with the type of food, with 4.5% of the protein in vegetables, 3.5% of the protein in fruits, and 10% of the protein in breads and starches consisting of leucine. Alternatively, 7% of the total protein from any food source can be assumed to be from leucine. For example, a slice of bread with 2 grams of protein will have an estimated 140 (7%) to 200 (10%) mg of leucine. Some clinics like to assume a larger percent, using a figure of 10%, while other clinics may assume less. It is important to check with your clinic for the appropriate calculations to estimate the leucine content of foods not on your list.

When illness or infection occurs, body proteins may be broken down. This can result in elevated levels of BCAA despite dietary compliance. It is a good idea to have a sick day protocol for treatment during illnesses. Frequently there will be a change in the medical formula recipe, and a reduction in the amount of leucine consumed from foods and in the formula. Increasing the amount of isoleucine and valine prescribed is typically indicated. It is always good to push fluids and calories to help flush out the toxins in the body. Additional medications, such as Zofran for vomiting, or Tylenol for fever, can help the individual, and it is always advisable to seek medical attention for other illnesses, i.e. antibiotics for ear infection. Parents are asked to monitor their child’s metabolic control during illness by checking the urine for ketones. It is important to keep the clinic informed of your child’s progress and to seek medical attention if there is no improvement. Use of an Emergency Care Protocol helps to ensure that the appropriate care is given promptly when acutely ill. The ER protocol should include information on the patient’s condition and the acute management needed along with contact information for the Metabolic team for further assistance.

Unfortunately, complications may occur. Feeding disorders may result from the anorexia and vomiting associated with compromised metabolic control. difficulties with enteral tolerance including reflux and other GI disorders, oral-motor difficulties, or organ dysfunction such as pancreatitis may also occur. Because of the restrictive nature of this diet, nutritional deficiency is a possibility. Deficiencies in trace metals, such as selenium and molybdenum have been reported as several of the formulas did not include them. Marginal calcium intake, along with the chronic acidosis related to elevated branched chain ketone levels, can lead to decreased bone mineralization and increasing the risk of developing osteoporosis. Many individuals following protein restricted diets for various inborn errors of metabolism have been found to be deficient in the essential fats, eicosapentanoic and docosahexanoic acids. Primary dietary sources for these essential fats are typically high in protein and excluded from diet. Ask your clinic about supplementation of these oils.

While we have focused on the need to restrict protein, problems can develop from over-restriction of protein as well. These include poor growth, fat deposits in muscle, decreased resting energy expenditure, skin rashes, decreased immunity, decreased appetite, hair loss, and osteopenia. Specific amino acid deficiencies can also occur such as acrodermatitis enteropathica associated with isoleucine deficiency. Valine deficiency can lead to poor...
growth, drowsiness, irritability, crying, and decreased blood protein levels. Thus it is important to ensure that individuals with MSUD are receiving sufficient amounts of all essential amino acids including isoleucine and valine while restricting leucine to the proper amount. Carnitine deficiency can cause fatty myopathy, cardiomyopathy, depressed liver function, and neurologic dysfunction, hence some individuals need to supplement this nutrient.

Dietary treatment changes with age. It is important to have regular follow ups with your Metabolic Clinic to make sure that the treatment is adjusted appropriately to account for growth and metabolic stability. It is important to comply with dietary treatment as the clinic will continually try to change the plan if it is not working, but the plan will not work appropriately if not followed correctly and communicated with the clinic.

There are many other factors that may have a large impact on dietary management for MSUD. Physical activity is very important as when individuals are more active, they develop more muscle. When they have more muscle, they can tolerate more leucine in their diet. I encourage all of our patients in our clinic to try to be as active as possible. Maintaining good dental hygiene and regularly seeing a dentist is important as infections in the gums and mouth can lead to compromised metabolic control. Traveling with MSUD can go well if planned in advance.

Treatments beyond diet are now being evaluated for MSUD, but these still need to be evaluated over the long term. In the future there may be medications that can help with management. Earlier detection and more successful treatment will improve outcomes. There will be more reports of successful pregnancies for women with MSUD, an increased experience with organ transplantation, and eventually gene therapy may be an option. But for the time being, dietary management remains the main treatment for MSUD.

Elsis Santos’ Quinceanera
By Zulma Santos (mother of Elsis)

The quinceanera is a young Hispanic woman’s celebration of her fifteenth birthday. This birthday is celebrated differently from other birthdays. The quinceanera is considered to be the most important birthday as it symbolizes the transition from childhood to womanhood. This event is usually celebrated with an elaborate ritual. Here is a description of Elsis’ quinceanera, which took place November 26, 2005.

Celebration of the quinceanera
An elaborate ritual is executed to celebrate the quinceanera. The celebration begins with a Thanksgiving mass (Misa de Acción de Gracías). At this mass, the quinceanera is accompanied by a head chamberlain (chambelan), several other chamberlains, maids of honor (damas), her parents, and godparents (padrinos). The quinceanera, chamberlains, and maids are all dressed in formal attire, but the quinceanera is uniquely dressed while the chamberlains and maids are in matching attire. Sometimes the head chamberlain is dressed uniquely as well to distinguish him from the rest of the chamberlains. At this mass, the quinceanera, along with the rest of her family and friends, thank God for all their blessings.

After the mass, a feast usually follows either at the family’s home or at a banquet hall. At this feast there is food and dancing. Special party favors (recuerdos) are distributed to the guests to remember this special event. During this feast a celebration dance is performed. It usually includes a choreographed waltz with the quinceanera, the chamberlains and maids of honor. In this waltz it is customary to have a portion dedicated to symbolizing the transition from childhood to womanhood. In this special dance there is also a portion dedicated for the quinceanera to thank her father, and other relatives, with a dance.

Elsis had to do a lot of work for this event, including photo shooting, dance practicing. It took a year to plan, and Elsis was cooperative and excited.

Elsis has classic MSUD. She was diagnosed when she was 9 days old and was really in a critical condition. She had to get dialysis and all the other medical interventions, and stayed in the Hospital for about 2 months.

Now Elsis is getting ready to go Forrest Park High School, 9th Grade. She enjoys playing games on the computer and enjoys her game boy. She also has a brother. His name is Anthony (2 years old) and she really enjoys helping take care of him. Anthony does not have MSUD. Even though Elsis did her Spanish celebration, now she is looking forward to her sweet 16th which is also part of her, because she was born in America, in Alexandria, VA.
**Improving Self-Esteem**
Darlene Honigford, LSW

Self-esteem is an issue that most of us struggle with. For a child with a chronic disease, the struggle is even greater. Darlene Honigford presented a clear discussion of what self-esteem is and how to nurture it in our children.

*What is self-esteem?*
Self-esteem is the collection of beliefs or feelings we have about ourselves. Experiencing a sense of accomplishment bolsters self-esteem, starting very early in life. It is about how much you feel valued, loved, accepted and thought well of by others, and how much you value, love and accept yourself.

Self-esteem is important as it gives one an immunity to the roller coaster of life. Rejections, disappointments and failure are a part of daily life. Life is not always fair or equitable and even our best efforts are not always successful. But high esteem can assist a person in “weathering the storm,” to look beyond immediate downward dips.

People with healthy self-esteem feel good about themselves, appreciate their own worth, have a positive attitude, take pride in their abilities, skills and accomplishments, see themselves as competent, and compare themselves favorably with others. Researchers have found that high self-esteem correlates highly with self-reported happiness.

Those who have a healthy self-esteem tend to enjoy interacting with others. When challenges arise, they are able to work towards finding solutions. These people are comfortable in social settings and enjoy group activities as well as individual pursuits. They know their strengths and weaknesses and accept them. They can voice discontent without belittling themselves or others, and have a prevailing sense of optimism.

Those with low self-esteem, on the other hand, feel as if no one likes them or accepts them. They feel they can’t do anything well, have a sense of powerlessness, and experience depression. They may not want to try new things and frequently speak negatively about themselves. They may exhibit a low tolerance for frustration, giving up easily or waiting for somebody else to take over. Easily disappointed in themselves, they see temporary setbacks as permanent, intolerable conditions.

Ones level of self-esteem has a great deal to do with how one reacts to having a chronic disease. Feelings of being different and, perhaps, damaged make it more difficult to feel good about oneself. The disease may cause frustration and anger. In fact, it often creates a paradox: The disease, MSUD in our case, must come first in your life, yet you don’t want it to take over your life. While following one’s treatment program is of utmost importance, there is a feeling that the condition has never been and never will be you. Coping with this paradox takes emotional honesty, maturity and support.

*Improving Self-Esteem*
A critical step is to stop thinking negative thoughts about yourself. To help accomplish this, the child should be encouraged every day to write down 3 things about themselves that make them happy. Focus on positive thoughts, not on thoughts of defeat or failure. The child should be encouraged to:

- Aim for accomplishments rather than perfection
- View mistakes as learning opportunities
- Try new things
- Recognize what you can change and what you can’t
- Set goals
- Take pride in your opinions and ideas
- Make a contribution ex: volunteer
- Exercise
- Have fun

Parents can help by:

- Watching what they say
- Praise and reward effort and completion of task instead of outcome
- Be a positive role model
- Nurture your own self-esteem
- Identify and redirect your child’s inaccurate beliefs
- Be spontaneous and affectionate with your child
- Give positive, accurate feedback
- Create a safe, nurturing home environment
- Make your home a safe haven for your family
- Help your child become involved in constructive experiences

If you suspect your child has low self-esteem, consider therapy to help your child adjust the way he or she views himself and the world.

Darlene closed with the following anonymous quote:

“Watch your thoughts, for they become words. Choose your words, for they become actions. Understand your actions, for they become habits. Study your habits, for they will become your character. Develop your character, for it becomes your destiny.”
News from Nutricia (formerly SHS) North America
Research Among Patients with Inborn Errors of Metabolism

Nutricia North America and SKIM Group are partnering to conduct research among patients with inborn errors of metabolism. The main objectives of the study are to:

- Learn about patient’s concerns regarding currently available clinical nutrition products
- Determine how clinical nutrition products can be improved

The research group is looking to interview 50 to 80 parents or adult patients with inborn errors of metabolism. The interviews will last about 30 minutes and will be conducted by phone. Participants in the research will receive an incentive for their time and effort.

SKIM Group, an independent third party firm, will be conducting the research and all information will remain anonymous. If you are interested in participating in this study, please e-mail Joyce Bentzman at jbentzman@shsna.com

Low in Protein, BIG in Taste!
Nutricia North America Introduces Milupa Line of Low Protein Foods

Nutricia North America is excited to announce the availability of new Milupa low protein (lp) foods that are low in protein and BIG in taste! These great tasting lp-foods are suitable for children and adults, provide on-the-go convenience and add variety to low protein diets. New offerings include:

- Drinks – unflavored and chocolate
- Snacks – paprika chips and italian mix
- Cereals – chocolate ringlets and flakes
- Cereal Mixes – pear and apple-banana
- Energy Bars – apple-cinnamon, apricot and fruits of the forest

Product information, recipes and online ordering are conveniently available at MySpecialDiet.com. Free shipping is available on orders over $25.

The following essay was written by David Fischler, a high school senior in Marietta, Georgia, as part of his college application.

To survive is to carry on despite hardships or trauma. It is an act which is second nature to humanity, but for the past eighteen years not only have I survived but I have prevailed over my hardships, my greatest personal accomplishment. At the age of sixteen days old I was diagnosed with a rare metabolic disorder known as Maple Syrup Urine Disease (MSUD), which affects the liver. Growing up in an age where little was known about this disease I managed to not only survive but to continue to surpass all expectations that were placed upon me. Over the past eighteen years, I have grown and continue to live a fulfilling life under many dietary restrictions and medications. These dietary and medical needs sometimes seem overwhelming. Also, people ask “What is that stuff you take?” or “Why can’t you eat pizza?”, but I have always and continue to fight through. With the support of my family, friends, and the Department of Genetics at Emory University, I have been privileged to live a life that only twenty years ago seemed unimaginable for anyone with Maple Syrup Urine Disease.

Due to this disorder it has been hard to participate in sports and many other outdoor activities. I dehydrate much faster with more severe consequences than everyone else. Furthermore, I can not get hydrated with water alone and I must drink sugar water, like Gatorade. However, I have been able to participate in activities which others take for granted, like soccer and most recently marching band. Through my protein restrictive diet and many medications, I am proud that I was able to play the sport of soccer for nearly fifteen years on a championship caliber team with my peers, and I continue to be a referee in the sport that I love. Also, I am glad that last year was the first year my doctors would allow me to join my high school’s marching band. I worked hard and felt a sense of achievement when the band won third place at the Band of America Super Regional in Atlanta. This year, I am once again in the band, as I try to continue having as normal a life as possible.

Over the past several years, I have grown to understand my medical condition, studied it, and met others with it. Through these interactions I have learned that I am one of the lucky ones. I could have very easily been mentally retarded by this disorder or worse, but I am not. I have survived. I have survived to live, what so far has been, a happy and fulfilling life surrounded by people who care about me. To live and grow up as well as I have and to not only survive, but prevail over this illness, is by far my greatest personal achievement.
Our MSUD Family Scrapbook

CONGRATULATIONS!

On August 19th, 2006, Michael Koons (25, Variant MSUD), brother to Matthew Koons (26, Variant MSUD both of Harrisburg, PA), married his fiance of 3 years, Jamie Fessler. They were married in Campbelltown, Pennsylvania and they had their honeymoon at Estes Park, Colorado, which is in the foothills of the Rocky Mountains. All had a great time and they are very excited about beginning their lives together.

The Pattersons (Oliver 12, MSUD variant and Eric 12) during our European vacation. 7 days biking tour (280 km) through the Czech Republic followed by 2.5 weeks of mountain hiking in Austria. We rented apartments or took a cooler with specialty foods from hotel to hotel. Oliver says that the potatoes in the Czech Republic are better than anywhere else!

To share accomplishments (large and small) with your MSUD Family, send photos with caption to: agcreative1@aol.com or Adrienne Geffen, 1025 Red Oak Drive, Cherry Hill, NJ 08003
Chex Mix
- 2 cups Rice Chex
- 2 cups Loprofin breakfast loops
- 1 cup Ener-G Food Wylde Pretzels
- 1 cup Loprofin snack Italian
- ½ cup small butter flavored crackers
- ¼ cup butter
- 1 T Worcestershire sauce
- ¾ teaspoon season salt
- ¼ teaspoon garlic powder
- ¼ teaspoon onion salt

Protein: 5.7 g
Leucine: 480 mg
Calories: 1174

Apple Dumplings
- 170 grams wheat starch
- 170 grams Cambrooke foods baking mix
- ¼ cup water
- ¼ cup liquid coffee creamer
- ½ cup brown sugar
- 1 ½ T Butter
- 1/8 teaspoon cinnamon
- 1 ½ teaspoon baking powder
- 5 baking apples

Protein: 4.32 g
Leucine: 304 mg
Calories: 3006

Chicken Fajita
- 1 Cambrooke tortilla wrap
- 1 Cambrooke Tweezk (chicken nugget)
- 1/3 cup sliced onions
- 1/3 cup sliced red and green peppers
- 2 tablespoons oil
- 3 tablespoons salsa
- 2 tablespoons Cambrooke cheddar shreds

Protein: 2.4 g
Leucine: 117 mg
Calories: 439
**NEW!**

**Cinnamon Crackels Cereal**
Crackels is a great tasting puffed tapioca cereal made available for the low protein diet from Applied Nutrition. They are available by mid September and you can order by going to www.dietforlife.com or phone 1-800-605-0410. Jordan and I both tasted them at the MSUD Conference and they are very good. Also available from Applied Nutrition is Chocquitos, chocolate raisins, white chocolate cranberries, protein free bars and Maddy's Tangles.

**NEW!**

**Cambrooke Buns & Tweekz**
Made just to fit the mushroom burgers from Cambrooke Foods these buns will give your child a great tasting burger that looks just like the rest of the family. Available in a pack of 8, each bun has .2 grams of protein, 22 mg Leucine and 130 calories. Tweeks are triangle chicken nuggets which are very tasty. There are 8-9 nuggets in a box; a serving of 3 nuggets is 1 gram of protein, 58 mg Leucine and 170 calories. These products can be ordered at www.cambrookefoods.com or by calling 1-866-4-low-pro.

Cambrooke has a very nice web store which is easy to access and all items are accompanied by photos. Also available are baking mixes, pastas, breads, baking mixes and many more items.

**NEW!**

**Wylde Pretzels**
Tasty little pretzel rings are now available from Ener-g Foods. One serving of 40 pretzels is 1 gram of protein, 48 mg Leucine and 130 calories. These are available in sesame, poppy seed and lightly salted. Ener-G Food can be reached by www.ener-g.com or by calling 1-800-331-5222. Also available are baking mixes, pasta, bread and various low protein products.

**NEW!**

**Taste Connections**
Taste Connections was a new company to me at the MSUD conference this past June. Available from them are cookies, popping tapioca, baking mixes and more. Taste Connections can be reached at www.tasteconnections.com or by calling 1-310-371-8861. There are recipes available on the web site for all many different kind of foods. Holiday cookies are now available in clear bags or in colorful tins.

**NEW!**

**Vitaflo Formula**
A new formula, MSUD express cooler in a convenient, ready-to-drink package is now available from Vitaflo. Also available are pre-measured packages of powder and flavor packets. Just add water, shake and drink. These are available at www.vitaflousa.com or 1-888-VITAFLO.

**Cambrerke Foods**
Tweekz™ are here - our new chicken-flavored nuggets! Just heat and serve these crispy nuggets for an easy, kid-pleasing meal. Camburger Buns are now available to complement low protein Camburgers – perfect for end-of-the-season picnics. To make any meal special, try our Italian Focaccia sticks.

Munchy Bites are back! The whole family will love these savory-flavored Sweet Barbecue Munchy Bites. These tapioca-based snacks have a great nutritional profile and are perfect for snacking or carrying to family events or in a school lunch. Request a School Lunch Program packet if you want your child to join the lunch line and enjoy a low protein lunch served by the school cafeteria. This packet will simplify the process for you and your school's food service staff.

Do you have storage questions or are you not sure how to stretch your low protein foods? Keep all frozen products (Tweekz™, fresh filled pastas and Camburgers) frozen. Prepare only what you need for each meal. Other products freeze well too, such as imitation cheeses, desserts and bread products.

Do you have storage questions or are you not sure how to stretch your low protein foods? Keep all frozen products (Tweekz™, fresh filled pastas and Camburgers) frozen. Prepare only what you need for each meal. Other products freeze well too, such as imitation cheeses, desserts and bread products. Only take out what you will eat each day. Breads thaw quickly and can be quickly thawed in the microwave wrapped with a white paper towel. Use products like flavored cream cheeses and sauce mixes to stretch other products and create simple low cost meals by using them as flavor enhancers and sauce bases.

Pasta go far when preparing meals and are very filling. Cook pasta, make a low protein sauce and add small amounts of low protein vegetables or meat alternatives.

We are ALWAYS open to serve you. Call toll-free, (866) 4 LOW PRO / (866) 456-9776 or visit our website at www.cambrookefoods.com. If this is not convenient, you can mail (2 Central Street, Framingham, MA 01701), e-mail (orders@cambrookefoods.com) or fax your orders to us at (978) 443-1318.

**Applied Nutrition** continues its commitment to provide low protein products with the look and taste of store bought foods. Crackels Cereal was specially formulated in the laboratories of Applied Nutrition to contain less than 12 mg of Leucine per 30 gram serving. Each box of Crackels contains 8-1 cup servings. Don’t miss the “Fun Stuff” games and puzzles on the back of every box. One cup serving of cereal has 120 calories, 0.2 grams of protein and 12 mg of leucine. Cinnamon Crunch Crackels looks and tastes just like your favorite store bought cereal but without the protein.

You can call or go onto the web site to receive your free 1 oz package of cereal.

Applied Nutrition is also the maker of Complex Vanilla formula.

For more information about Crackels, Tangles chips or any low protein products brought to you by Applied Nutrition visit www.DietForLife.com.
A FAMILY HISTORY

Connor Patrick McMahon

Our son Connor Patrick McMahon was born on St. Patrick's Day in 1996. My wife and I are both Irish, so it was a dream come true for both of us. On Connor's fourth or fifth day of life he stopped drinking. His eyes closed and remained closed for several days. Through the Newborn Screening test we were made aware that Connor had Classic MSUD. Like most of you reading this, we were devastated when we heard the news.

Connor's first few years of life were spent with frequent hospitalizations due to elevated leucine levels, mostly caused from illness. From ages one to five Connor experienced a few ICU visits. Although to have him in ICU is a big scare, it is the best place to have him when he is ill. Thankfully, he was able to overcome each episode he was dealt.

I feel like I have to make you aware of some of the darker moments we have had to deal with, in order to fully appreciate the brighter ones. There are so many bright moments in Connor's life that we feel like we are bragging about him, but here are some anyway.

Connor is a bright, athletic child with a wonderful sense of humor. He is a straight A student and accelerates in math. In our Spring teacher conference this past June, we were told that Connor is the class leader and that he is such a role model to the other children. Connor has always been discrete about his disease. I think some of his classmates might not even know he has it. It is just something that he deals with and he feels no different. Connor plays hockey, baseball & football on various organized teams. He excels in all these sports and enjoys them very much. Connor's disease has not affected his ability to excel in athletics. Connor is a very popular child with several different groups of friends. He is very social and very active.

Connor is able to tolerate 1,200 mg/leucine daily. His daily diet consists of mainly cereal, low pro bread, pasta, fruits & some vegetables, snacks of all sorts & potatoes, potatoes, potatoes!!

Connor is tall & thin. He is now ten years old, 5' tall and weighs 76lbs. Connor has had some difficult times dealing with his disease, but for the most part he is acceptant and understanding of what he is faced with.

I'm sure there may be some difficult times ahead, but with the diligent work from his mother and the strength of Connor we will get through them. God has blessed Connor with so many wonderful qualities that it has made it a bit easier to overcome things.

While many people may look to athletic superstars, business executives, priests, and doctors for their inspiration, our son Connor is our true inspiration. The way he deals with his everyday diet, the way he may not always feel his best but continues to carry on and live a normal and wonderful life. Thank you Connor for inspiring me, your mother, your sister and hopefully many others who deal with what you have to deal with.

NEWS FROM ROSS METABOLICS

We have recently revised A GUIDE FOR THE FAMILY OF THE CHILD WITH MAPLE SYRUP URINE DISEASE. It is available in both English and Spanish. To obtain copies, please ask your nutritionist or call Ross Metabolics at 1-800-986-8755 (option 1). Additional copies of the Spanish MSUD Family Guide can also be obtained by contacting Monica Falconer, 10601 Washington Cir., Anchorage, AK 99515.

ROSS -
Providing specialized nutrition for infants, children and adults with inborn errors of metabolism.

The trusted source of Ketonix® -1 and 2.
My daughter, Libby (age 21), and I attended the symposium in Ohio. Our family has been thinking about and gathering information concerning the liver transplant for the past two years, and we were excited to learn that this was going to be a main focus of the symposium. We, as a family, have been praying that God would help us to make the right decision about going forward with the liver transplant. When Dr. Morton spoke that last day, Libby and I both felt immediately, that this was the answer we had been waiting for. We have since moved forward, and Libby had her liver transplant evaluation the first week of August. Our family is so excited that Libby will soon have a new lease on life, but are certainly aware that our blessings will also be someone else's loss.

We would like to thank everyone involved in helping with the symposium. We really appreciate all the time and effort it takes for you to make this happen. It truly was one of the best. The accommodations were excellent and the information was outstanding. Having the extra time to spend visiting with the other families has always been a rewarding time. We have been attending every symposium since the one held in Montreal, and our bond as an MSUD family strengthens each and every year. We will be looking forward to seeing all of you again in 2008.

Robin and Libby Stone, Desoto, KD

Recently I was contacted by a friend of the family and neighbor of mine, Charles Giuliano who had an on-line magazine called Maverick Arts out of Boston. In the e-mail and phone call Charles told me that he was going to start a Berkshire branch of his magazine with its own website. Since I enjoy watching plays and movies, Charles asked me if I would be interested in writing reviews for some of the summer plays and films that were performed or had viewings in the Berkshires. This included the Williamstown Theatre Festival, The Berkshire Theater Festival, and Barrington Stage. I watched a couple documentaries at a small movie theater in Williamstown that gets a lot of the independent and foreign films, called Images Cinema that Christopher Reeve used to endorse.

The reviews I wrote for Maverick Arts were for a production of “Anything Goes,” starring Sharon Lawrence (best known for her role on NYPD BLUE), one on two documentaries, “Word Play” (about the New York Times Crossword Puzzle and its National tournament) as well as “An Inconvenient Truth” (Al Gore’s documentary about his mission to educate people about global warming and its effects on the earth). The third article was about the upcoming plays that were going to be performed this season on the main stage of the Williamstown Theatre Festival.

I’m finishing up two reviews on Romeo and Juliet, where I met and interviewed Emmy Rossum and the guy that played Romeo for the review and “Night of the Iguana” which starred Linda Hamilton (best known for the Terminator 1 & 2 movies).

Emmy’s performance as Juliet made the show. Unfortunately, since my camera broke after interviewing Austin, who played Romeo, I asked if it would be alright with her if I were to come back later to take a picture for her quote. A week later I ran into her after the play. However, Emmy didn’t want to have her picture taken alone, so she asked me to be in it with her since I did the interview. She also signed the cover of my “Phantom of the Opera” movie. She was extremely nice and charming and we hope to bump into each other again.

-- Nikolai Rudd
16th Annual Auction
to benefit Clinic for Special Children

Each year thousands gather at the Leola Produce Auction and bid on donated items, raising what amounts to 1/3 of the Clinic’s yearly operating budget. Homemade items, furniture, quilts, wallhangings, farm equipment - even a piano(!) can be found on any of the auction stages. The talented and entertaining professional auctioneers all volunteer their time, as well, to benefit the clinic. Drs. Morton and Strauss each gave a heartwarming talk about the clinic and their appreciation to the Mennonite and Amish Community coming together to make the auction a success which allows them to carry out their most important work.

Mark your calendar for the 3rd Saturday in September (Sept. 16, 2006) for the 16th Annual Auction, an experience to be remembered!!

The Leola Produce Auction is located on Brethren Church Road, north off Rt. 23 in Leola (between Lancaster and New Holland). Call 717-626-4863 for more information.

Jimena Gatica
Buenos Aires - Argentina

Jimena received a partial liver transplant on July 5, 2006. She was the first MSUD child to be transplanted outside of the States, and the first in the world with the reduced liver technique. Her story appeared in local newspapers at the time.

The partial liver technique consists of putting a lobe of a donor’s liver in place of the old liver. Then the liver grows to fill the entire cavity. As liver is trimmed it takes more time to recover than a whole liver, but complications are the same for both procedures. The transplant team at Hospital Italiano of Buenos Aires is very experienced in this technique. They have done more than 200 transplants.

Jimena’s case was complicated by fluid in her right lung. She was discharged 54 days after transplant.

Jimena’s experience is important for children with MSUD all around the world. In Argentina, as well as many other countries, the lack of pediatric donors makes it very difficult to wait for a whole organ that matches exactly. The reduced liver technique reduces waiting periods in countries like Argentina.

I received a lot of emails from parents of children with MSUD from Brazil, Chile, Mexico, Honduras, New Zealand, etc that are interested in knowing more about the procedure.

Eduardo Gatica
Father of Jimena - 2 1/2 years
MSUD Transplant Symposium

Patients and families reunited with doctors and other caregivers at Children’s Hospital of Pittsburgh’s (CHP) Second Annual Maple Syrup Urine Disease Symposium held on May 19th, 2006 at the Wyndham Garden Hotel in Pittsburgh.

Several physicians made presentations about advancements in the treatment of MSUD patients. In 2004, CHP developed the world’s first protocol for performing liver transplants in MSUD patients. This protocol led to 18 patients receiving liver transplants at Children’s, the most of any center in the world. CHP’s comprehensive protocol includes the development of specialized computerized order sets (which direct the transplant team every step of the process); the establishment of a completely sterile room needed to mix amino acids for MSUD patients in the event of a metabolic crisis; and specific formulas called total parenteral nutrition (TPN) formulas in the event of a metabolic crisis.

More than 70 people attended the symposium, including many transplanted MSUD patients and their families, caregivers from CHP, the Clinic for Special Children in Strasburg, Pa., and the University of California San Diego School of Medicine. Presentations were made by: George V. Mazariegos, MD, director of Pediatric Transplantation; Gerard Vockley, MD, PhD, chief of Medical Genetics; Hilary Feldman, PhD, of the Child Development Unit, Kevin Strauss, MD of the Clinic for Special Children and Ajai Khanna, MD, of University of California, San Diego. Time was set aside for questions and answers following each presentation.

Physicians from Children’s and the Clinic for Special Children in Strasburg, Pa. reported on results of liver transplants for Children’s first 11 MSUD patients in April 2006 in the American Journal of Transplantation. The patients’ amino acid levels stabilized within six–12 hours of transplant and have remained stable since despite unrestricted intake of protein.

Elan Geffen, transplanted 1/17/06, celebrated his 22nd birthday with friends while attending the MSUD Transplant Symposium.

Poems by Nikolai Rudd

SILENT CHORDS
Now, in loud percussion
Heralds the music’s silence.
But still sweetly plays in mind.

The violin sings its sad tale,
The cello plucks its deepened wail.
The bass, filling in sporadic chords.

From composers long since dead,
Plays their music inside my head.
The waves of emotion roll like hills—
Their genius shows those deadened skills.

Long have we listened—
The single notes, strung together—
Give resonance to their form.

BEAUTY
What fascination has beauty?
’Twas the Bard’s proclamation—
“Beauty too rich, for words too dear.”
That in semblances, transfixed
For it can, and does entangle those within its stare.

There are words—
But none that can, but try
Unfathomable! Beyond all grasp
Feelings that arise from nowhere
Stir the jolted heart’s clasp.

A richness that calls us all.
Some go running, while others crawl.
A perception of the mind.
Dangling above the beating heart.
Forever, a journey’s pride.
A Note From the Editor

This issue of our newsletter focuses on the Symposium, held June 15-17 in Dublin, Ohio and hosted by the Bulchers. Many thanks to Sandy and Dave Bulcher for all their hard work. Like the newsletter, the Symposium provides a link for those of us who often feel alone. We relish the opportunity to meet each other, for our children to connect with others going through similar challenges, and to become energized by learning about all of the advances being made in the treatment of MSUD. Without the Bulchers, this would not have happened.

As you will see by reading the articles in this newsletter, we learned about promising advances in both gene therapy and liver transplantation. Sessions on dietary management and self esteem provided us with many of the coping skills that we need on a daily basis.

My daughter Hannah (age 12) and I had to leave before the family gathering at Coffman Park as she was attending the Metabolic Camp at Emory for the first time. Hannah had quite a summer. After making new friends and gaining confidence at both the symposium and Metabolic Camp, she spent 2 weeks at sleep-away camp for the first time. The camp was an hour away from our home and about 1.5 hours from Mt. Sinai Hospital where she is treated, allowing her Dad and I to feel comfortable knowing that we could act quickly should she become ill. We delivered a weeks worth of formula with her and another weeks worth when we came for visiting day the following weekend. With the help of her counselor, she responsibly measured her food and kept track of her leucine intake. Not only did she have a wonderful time, but her subsequent blood test showed perfect amino acid levels. Success!

Several families and individuals have shared pieces of their lives with us in this newsletter. We hope you will all consider doing so. Each person's story is valuable to us. Send submissions to me at krdhed@aol.com.

As I'm sure you know, it takes more than one person to prepare a newsletter. Special thanks to Adrienne Geffen, who lays out the articles in preparation for printing. Believe me, this is a very time consuming job.

Have an enjoyable fall and holiday season. We'll talk again in January through our Winter issue.

Karen Dolins
Mom to Hannah – Age 12

Symposium 2006 Attendees said...

Emily Anderson- MSUD mom from IL “Meeting families with other young MSUD children who are healthy and happy was my favorite part of the symposium-it was reassuring and inspiring. We so enjoyed, too, hearing about the latest research and treatment.”

Elizabeth Emerick- RD from MD “This was a wonderful conference. I think that this was the best conference yet. There was alot of opportunity for informal interaction.”

Paula Ruter- MSUD mom from MI “When we are at home we feel isolated. It means the world to talk to parents that actually know what we are talking about.”

MSUD parent: “We enjoyed the information regarding liver transplants and progress on the mouse model.”

Julie Szymczak- MSUD mom from IL “This was our 3rd symposium and we learn each time that we attend. I have to say that we learn the most from talking with other parents. We enjoy every time that we attend.”

MSUD parent: “It always is a pleasure to meet with other families with the same concerns and problems. It has been a blessing to attend again.”