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MEDICAL/NUTRITIONAL CARE PLAN

Details

Written by Keiko Ueda, MPH, RD, LD

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Keiko Ueda is a dietitian with the Division of Genetics/Metabolism at the Boston Floating Hospital for Children at the New England Medical Center in Massachusetts. She spoke to the MSUD families on Practical Nutritional Considerations in MSUD at the National Coalition For PKU and Allied Disorders Metabolic Conference *in May 2001. In this article, Keiko focuses on the support systems needed by most families in order to provide adequate nutrition for those with MSUD.*

Keiko Ueda and Shayla Brubacher at the National Coalition Conference, May 2001

Introduction

Decisions regarding the medical/nutritional care plan for a child or adult diagnosed with MSUD is a team effort - with parents and caregivers often expected to take on the role of team leaders. Input from the patient, parents, and caregivers is required, most of all, in order to meet the daily medical and nutritional needs of a MSUD patient. Therefore, ongoing communication with your primary care physician, metabolic physician, and metabolic clinic staff is key. Keeping the lines of communication open with your metabolic clinic will best assist them in helping you tailor the MSUD diet to your child's needs and adjust the diet as medically necessary to optimize metabolic control, growth and development, and maintain

and support good health. If there are multiple daily caregivers (parents, relatives, teachers, nurses, sitters, etc.), it is important for everyone to be consistent and aware of the most current MSUD nutritional plan in order to avoid errors or mis- communication.

I have great respect and admiration for the dedication and daily efforts of the patients, parents, and caregivers of individuals living with MSUD and other metabolic disorders. As we all know, coordinating the medical nutrition therapy for an individual diagnosed with a metabolic disorder such as MSUD is a challenge. As a metabolic dietitian, whenever I am planning the metabolic nutrition diet orders for an individual patient, I try to take into consideration individual, age-specific nutritional needs, as well as the practical issues that might influence or hinder the actual achievement of the goal dietary intake. I always try to consider each individual's short and long term dietary requirements and goals. I try to target metabolic education nutrition counseling to the needs and abilities of each individual living with MSUD. It is important to strive for realistic and achievable goals to avoid unrealistic expectations that can result in frustration and disappointment. Obviously, MSUD patients and their nutritional needs and goals vary greatly from one individual to the other. In my experience, I have found that MSUD patients have distinct nutrition and feeding issues and needs at different times and stages in their lives. Most of the MSUD patients I have known do well meeting their daily nutritional needs via oral feedings when in good health. During times of illness, they often require the support of tube feedings (TF) and/or intravenous (IV) therapy to assist in illness recovery and regain metabolic control.

MSUD dietary goals and restrictions can be very different between individual MSUD patients, and for each MSUD patient in different medical situations at different times of his or her life. An individual's MSUD diet plan depends upon his or her diet tolerance; medical and nutritional needs for growth, development, and/or health maintenance; biochemical lab results; and medical/illness status. I would urge you to continue to discuss your individual questions and concerns with your metabolic clinic physician, dietitian and staff.

Overview
There are certain practical considerations that must be addressed in order to optimize a MSUD medical nutrition therapy plan to ensure the best possible outcome.

MSUD Nutrition Practical Considerations - Availability ofResources/Tools

- State Department of Health Newborn Screening Program for MSUD
- Metabolic physician services and staff
- Health insurance coverage/state legislation
- State Department of Health assistance programs
- MSUD medical foods (formula) & low protein specialty foods
- Supplies: digital gram scales, books (leu/protein food counts, low pro cookbooks)
- L-amino acid medication supplements (l-valine, l-isoleucine)
- Local or home outpatient lab draws and home monitoring (blood amino acid levels and/or urine DNPH)
- Emergency illness management protocols
- Support for MSUD diet at school (and/or work)

The ongoing availability of these medical services and nutritional supplies is important - these are the tools that will assist you in implementing the MSUD diet on a daily basis. I

have found that patients who do not have continuing access or have only limited access to these resources often have difficulty adhering to MSUD diet goals.

Calling Your Health Insurance Company
Medical health insurance coverage of MSUD Medical Foods (formulas) and low protein specialty foods are often key issues. If you haven't yet had difficulties with your health insurance regarding coverage of formula and/or low protein specialty foods, unfortunately chances are high that you may encounter some problems in the future. It is very important to understand your health insurance coverage plan benefits, policies, and limitations.

Health Insurance Coverage - When Calling Your Insurance Company

- Try to be patient and courteous, inform and educate, try to gain an advocate
- Review your health insurance plan coverage benefits and limits
- Document each call and contact person, info, department, phone numbers (extension), date of call
- Request information in writing for future reference
- Request a copy of your Explanation of Benefits (EOB)
- Request case management services if available to assist in care coordination
- Refer to MSUD formulas as "MSUD Medical Food," prescribed by your metabolic MD
- Know your state law and its limits (if available) and have a copy in your files

Your need to be diligent does not end even when you receive approval for health insurance coverage. Some health insurance providers require periodic reassessments and regular prior authorizations for coverage of MSUD Medical Foods, low protein specialty foods, and certain medications. Ask if your authorization has an expiration date in order to be prepared to renew it in the future - and then keep track in your calendar to allow for enough time (4 weeks or more) for the renewal process. Ask if your MSUD Medical Foods are covered under your health benefits, prescription benefits or durable medical equipment benefits. Your policy coverage may vary, have limitations, or have different co-pays or deductibles depending upon the type of coverage under which your MSUD Medical foods, low protein specialty foods, and amino acid supplements are placed by your health insurance company.

Who Will Supply Your MSUD Medical Foods and Low Protein Specialty Foods?

MSUD Medical Foods
You may find it best to order your MSUD Medical Food from a local pharmacy and make the monthly trips to pick it up. This is convenient (as long as the pharmacy can do direct insurance billing), sparing you the need to pay out of pocket, submit insurance claims with your receipt, and wait for reimbursement. Depending upon your insurance policy and state laws (if applicable), you may have out of pocket co-pays or deductibles for your MSUD Medical Food prescriptions.

Some families fill their MSUD Medical Food prescriptions via home health infusion (HHI) or durable medical equipment (DME) companies contracted with their insurance and have delivery services to their homes. This option depends upon your health insurance benefits and coverage. Insurance companies often contract with different home health companies, therefore you may want to ask for a list of providers contracted with your insurance company. In most cases, your metabolic clinic staff must contact the HHI/DME to provide

prescriptions and refill authorizations and to set up the initial deliveries to your home. Often the HHI or DME companies have limitations in terms of what they can provide. Most contracted HHI or DME companies can order the metabolic medical foods and bill your insurance. Some companies are limited by company policies or state license to provide only "enteral feeding supplies" (meaning supplies necessary to support a patient getting their metabolic medical food by nasogastric or gastric tube feedings). Home health infusion companies are licensed pharmacies, and are able to assist patients who require IV medications or nutrition support. Some HHI or DME companies can not assist patients who are taking all of their formula by mouth because of insurance or company policy limitations. Your metabolic clinic staff should be able to help you sort out the best options for your needs and family.

Health Insurance Coverage Issues - Denials

- Find out WHY? Request it in writing.
- Find out how to appeal: how long it takes, how/who will get response
- Request your metabolic clinic to provide a Letter of Medical Necessity
- Find out if you are eligible for your state Medicaid insurance coverage (or as a secondary provider)
- Find out if you are eligible for your state Department of Health assistance programs
- Contact your state Department of Insurance to check if your health insurance is in compliance with state law
- Ask your benefits office if your insurance is a "self funded plan" exempting them from following state insurance mandates (ERISA) if you have a state law
- Contact your local and state legislators for assistance
- Consider obtaining assistance from an advocate
- Consider changing your health insurance provider (make sure coverage is better, for example, watch for pre-existing condition exemptions)

Low Protein Specialty Foods

In terms of the low protein specialty foods, some state laws specify limitations of coverage per year. For example, the Massachusetts state law sets a maximum of $2500 per year per patient for low protein specialty food health insurance coverage. Most families direct order and pay the low protein food companies, then submit claims to their health insurance and wait for reimbursement. It is very rare to find a pharmacy, HHI, or DME company willing to order and successfully bill health insurance companies directly for the low protein specialty foods. In some cases, depending upon their insurance policies, families are able to arrange for the low protein food companies to submit bills directly to health insurance companies. But not all low protein food companies provide this service. This is a very convenient service, if available, but typically requires many calls and a lot of time spent filling out forms to put this in place. However, caregivers often find it worth the effort.

If your health insurance offers case management with an assigned caseworker (usually a nurse) or if you can find a helpful member services contact person to assist you with your insurance coverage issues, this can save you some time and effort in the future.

Finally, some states have department of health sponsored programs to assist families with obtaining metabolic medical foods and/or low protein specialty foods, for example: Massachusetts - PKU and Other Metabolic Disorders Special Dietary program; Maine - Children with Special Health Needs program; Illinois - Division of Specialized Care for Children program; your own state - Women, Infants and Children (WIC) program. Most

programs have eligibility requirements: state residency, diagnosis, financial need, written insurance denials for coverage, and/or patient's age. Discuss with your metabolic clinic staff whether there is a program in your state to assist you.

What to Do If Your Insurance Company Denies Coverage

Should you get insurance denials for coverage, you can research the appeals process for your insurance company. You can request assistance from your metabolic clinic staff; their phone calls and letters of medical necessity can support the need for a more in-depth insurance medical review. This can be a frustrating process that may take some time for review and decisions. It is important to continue to be as patient and courteous as you can in your interactions with your insurance company. Try to consider your correspondence with your insurance company as an opportunity to educate and inform insurance workers about MSUD and the strict and difficult medical nutrition therapy requirements for patients. By outlining just what you and your family have to go through (rather than taking a belligerent approach), you might just gain an ally at your insurance company who is willing to assist you in finding a solution to these (and other) problems and concerns.

Appendix I is a *sample* medical necessity letter that may assist you in your insurance appeals process. You might need the assistance of a legal advocate to support your appeals if you continue to get illegal denials for coverage in spite of all your best efforts. If your employer offers a "self funded insurance plan," the ERISA (Employee Retirement Income Security Act) exemption poses an obstacle to getting coverage; this apparently exempts the employer/health insurance from having to follow certain state insurance mandates. You must then appeal directly to your employer (through the benefits office) and/or insurance company on the basis of medical necessity for assistance with coverage. A few families have had success with the support of local or state legislators in reversing insurance coverage denials, even without a state law. But if your state doesn't currently have a state law mandating health insurance coverage for medical foods or low protein specialty foods, contact your local state and national parent support groups to assist in organizing grassroots efforts to get a law passed. (This has been successful in several states.) Providing your legislators inarguable information on medical necessity and financial burden of medical costs without insurance assistance will often win political support and sponsorship of a bill. If all else fails, it may be necessary to look for a different insurance provider.

If changing your insurance carrier is a consideration, make sure to do your homework to check the benefits of the new plan. Watch out for pre-existing condition exemptions, and ask how they would provide for your MSUD medical needs before finalizing the change. The federal law, Health Insurance Portability and Accountability Act (HIPAA) of 1996 (sometimes called "Kennedy-Kassebaum") might be helpful to continue to obtain coverage and avoid pre-existing conditions clauses (Reference 1). It is also important to know if your health insurance plan sets age limits (e.g., 18 yrs.) for coverage of dependent children. "Given the wide variation among state laws and individual policies, it is recommended to contact your insurance company or plan sponsor at least several months before the child turns age 19 to tell them that they have a dependent adult child with a disability. . . [it is critical to obtain state-specific advice]."[1]

Getting Organized

It takes much of your time and effort to have all of your various medical services organized, insurance coverage verified, and in place. I would recommend a methodical and step-wise approach to finding solutions to challenges that you may face in order to ensure that your child obtains the medical and nutritional support and services needed to promote good MSUD metabolic control.

Parents and caregivers of individuals with special health care needs are now expected to take on increased responsibility for coordinating their child's care. Caregivers often must work with multiple institutions and agencies: primary care physicians and staff, specialty physicians and staff, hospitals, outpatient labs, state department of health programs and case workers, health insurance company employees, home health supply companies, durable medical supply companies, pharmacists and staff, visiting nurses, etc. It can be overwhelming for many caregivers and patients to organize all of the contacts and information. Therefore a MSUD Individual Health Plan (IHP)[2] should be included in your records, along with your MSUD emergency illness protocol (see Appendix II). The IHP may help both caregivers and medical and specialty physicians and staff in organizing and recording important contacts and medical/nutrition issues. However, the IHP requires regular updating.

Summary

Staying in touch with your metabolic clinic physician, dietitian, and staff provides the best assistance as you adjust MSUD medical nutrition therapy goals for your child and his or her changing needs over time. Obtaining and maintaining health insurance coverage of MSUD Medical Foods, low protein specialty foods, and L-amino acid medication supplements can be very complicated. It is important to know the benefits and limitations of your health insurance policy, what your state law allows, and the availability of other state program resources. Ask your primary care physician and your metabolic clinic physician and staff for information to help you decide which resources are most helpful for your situation. Coordination of your child's MSUD medical and nutritional care can be one of your biggest challenges; organizing your information and contacts can greatly assist you and save your time.

Note from Keiko Ueda : I would like to thank Dr. Mark Korson for reviewing this article, and most of all, my thanks to the parents and individuals living with MSUD with whom I have been privileged to work and from whom I have learned the most.

***References:**

1. Schulzinger, R. 2000. *Youth with Disabilities in Transition: Health Insurance Options and Obstacles.* An occasional policy brief of the Institute for Child Health Policy, Gainesville, FL (ISBN: 0-9700909-4-3) or (www.mchbhrtw.org/materials)
2. Silva TJ, Sofis LA, and Palfrey JS. 2000. *Practicing Comprehensive Care:* A physician's operations manual for Implementing a Medical Home for Children with Special Health Care Needs. Boston, MA: Institute for Community inclusion/UAP, Boston.

Appendix I: Health Insurance Letter of Medical Necessity for MSUD

(*Sample letter - change all underlined sections to fit your individual case*.)

January 3, 2002
Attn: Medical Appeals Review
Health Insurance Company

Phone # and Fax #
Re: A.B. DOB: 12-20-98 *(A.B.: replace with name of individual; DOB: insert date of birth)*
Insurance ID #123456789

To Whom It May Concern:

This is a letter of medical necessity for A.B. to receive coverage for MSUD Medical Food manufactured by MSUD Medical Food (formula) company, low protein specialty foods from low protein specialty foods company(ies), and L-valine and L-isoleucine amino acid powders by the amino acid manufacturer company. A.B. has been diagnosed with a metabolic disorder called Maple Syrup Urine Disease (MSUD).

Medical treatment of MSUD patients requires a strict dietary restriction of the three branched chain amino acids - L-leucine, L-isoleucine and L-valine - because of the metabolic enzyme impairment in the branched-chain alpha ketoacid dehydrogenase (BCKAD) complex. A.B.'s daily intake of these three essential amino acids is restricted to the minimum amount he needs to promote continued growth and development, while also preventing any metabolic imbalance. It is also very important to try to avoid catabolism by ensuring daily intake of adequate total calories and providing enough of the other essential amino acids. The MSUD Medical Food is specially formulated to provide trace amounts of the three amino acids (L-leucine, L-isoleucine and L-valine), adequate amounts of other essential amino acids, as well as carbohydrates, fat and vitamins and minerals.

To elaborate, A.B. requires a diet leucine (therefore protein) food restriction of 530 mg leucine per day in order to control his blood levels to treat his MSUD. This means that he is only allowed a total of about 7 grams of protein from foods per day. This protein restriction would be equivalent to about 3 slices of regular bread, which obviously as a total diet intake of food per day would not be sufficient to meet his daily nutritional needs. This means that in order to continue meeting his daily protein and calorie needs, while lowering his dietary leucine intake, he needs to daily consume a prescribed amount of the MSUD Medical Food and also consume low protein (low leucine) specialty food products. Some examples of low protein specialty food products A.B. needs are: low protein pastas (e.g. 1/4 cup of low protein pastas contain about 0.1 grams of protein vs 1/4 cup regular pasta contain about 3.0 grams of protein); and his parents make low protein breads (1 slice low protein bread contains about 0.2 grams of protein vs. 1 slice of regular bread which contains about 2.5 grams of protein). These low protein specialty food products are not widely available in local supermarkets and must often be obtained from mail order specialty food companies.

The MSUD Medical Food, low protein specialty foods, and amino acid powder currently provides about 80% of A.B.'s daily nutritional needs for treatment of his disorder. The L-valine and L-isoleucine amino acid powder is prescribed when it is necessary to help meet A.B.'s daily minimum requirement of L-valine and L-isoleucine without concomitantly increasing his intake of L-leucine. Treatment for classic MSUD patients without the use of the MSUD Medical Foods, low protein specialty foods, and L-amino acids would result in protein-calorie malnutrition, metabolic imbalance, delayed growth, delayed development, neurologic impairment and possibly even death.

We appreciate your assistance in the coverage of the MSUD Medical Food, low protein specialty foods, and L-valine and L-isoleucine amino acid powders that are essential to

A.B.'s continued medical treatment of MSUD. As you know, coverage of MSUD Medical Food, and low protein specialty foods are now mandated in the Commonwealth of Massachusetts by the passage of MA House Bill 5622 signed into law by Governor William Weld on January 4, 1994. With appropriate treatment, we are confident that A.B. will achieve his growth and development potential within the limits of MSUD. We request your approval for coverage of his MSUD Medical food, low protein specialty foods, and L-valine and L-isoleucine amino acid powder, which are essential to continuing his MSUD treatment and promoting continued growth and survival.

If you have any questions regarding any of this information, please do not hesitate to contact us. We would appreciate it if you would inform us of your decision on this matter as soon as possible. Thank you for your time and consideration.

Sincerely,
Metabolic Physician, M.D. Metabolic Dietitian, R.D.
Division of Metabolism Division of Metabolism
cc: A.B.'s caregivers (list by full name)

Appendix II: MSUD Individual Health Plan
[Form adapted from *Silva, et al, 2000. Practicing Comprehensive Care.*]
(*Sample form - insert your information, add lines and adjust to your needs - examples are in italics.*)

Date of Last Revision: *1/3/02*
Name of Patient:
Date of Birth:
Caregivers Name(s):
Address/Phone #:
Emergency Contact: name and phone #:
Primary Care MD:
Clinic Address:
Phone #/Fax #/ After hours contact #:
Metabolic MD:
Hospital/Clinic Address:
Phone #/Fax #/ After hours contact #:
Principal Diagnosis: *MSUD*
Insurance Company and ID #:
Insurance Member Services Phone #:
Insurance Caseworker Name & Phone #:
Concerns for Upcoming Visits/Follow up Issues List:
1.
2.
3.

Consultants/Hospital/Phone #/ Last Seen:
1. *Dr Jones, Metabolism, Hospital, #, 12-2-01*
2. *Dr Smith, Neurology, Hospital, #, 11-5-01*
3.

Admissions History (last 12 months):
2/1/01 for a virus

Medications: Name/How Much/Schedule:
1. *L-valine/ 10mg/ml solution/4 cc twice a day*
2.
3.

Allergies:
Immunizations/Dates:
Home Medical Supplies/Where Obtained/Last Refill Date:
1. *MSUD Medical Food/home health care company/12/2/01*
2. *L-valine solution/pharmacy/12/2/01*
3. *Low protein specialty food order/company/12/2/01*

Contacts: Names and #s:
Home Health Company/DME:
Visiting Home Nurse:
Department of Public Health Program(s):
Early Intervention Caseworker:
School/Address:
School RN:
EIP/School Contact:
Pharmacy:
Respite Care Contact:
MSUD Nutrition Goals:
MSUD Formula Recipe:
MSUD 24 hr Formula Goals:
MSUD Formula Feeding Schedule:
MSUD Leucine (mg) or Protein (g) Food Restriction Goals:
Lab Testing Schedule/Date for Next Test(s):
1. *Blood amino acid levels in 2 weeks: 1-16-02*
2. *Home urine DNPH as needed*

Caregiver/Patient Goals for the Next 12 Months:
1.
2.
3.

NEW INSIGHTS INTO THE CAUSES OF BRAIN DAMAGE IN MSUD

Details

Written by Dr. Huseyin Mehmet

Published: 20 July 2009

Dr. Huseyin Mehmet (London, England) is a lecturer in neurobiology at the Weston Laboratory, a division of Paediatrics, Obstetrics and Gynecology at the Imperial College of Science, Technology and Medicine, Hammersmith Hospital. He is doing research which may eventually have an important impact on infants with MSUD. He will be sharing more on his research as a speaker at Symposium 2002.

Maple syrup urine disease (MSUD) is an inborn error of metabolism caused by a deficiency in branched chain a-ketoacid dehydrogenase, leading to the accumulation of the branched chain amino-acids (BCAA), leucine, valine and isoleucine, and a corresponding increase in their a-keto-acid derivatives (BCKA), a-keto isocaproic acid, a-keto valeric acid, and a-keto-?-methyl-n-valeric acid levels. Acute neurological deterioration in children is often associated with increased plasma and cerebrospinal fluid (CSF) concentrations of BCAA and BCKA. Magnetic resonance imaging studies in MSUD children have confirmed extensive white matter loss and neuronal injury.

Although the underlying mechanisms of cellular toxicity are not known, there is direct evidence that BCKA affect mitochondrial enzymes resulting in impaired energy metabolism. It has recently been demonstrated by a number of laboratories, including our own, that reduced mitochondrial function can trigger cell death by apoptosis (a well conserved and highly regulated mechanism of cell death used for the removal of unnecessary, surplus, aged or damaged cells). From a therapeutic point, apoptosis is attractive since it can be inhibited at specific stages, in some cases resulting in tissue recovery. Since concentrations of BCAA are increased in the CSF, we hypothesized that pathological changes in the central nervous system of MSUD children may reflect a neurotoxic effect of BCAA and BCKA triggering inappropriate apoptosis of neural cells.

Our results (recently published in *Molecular Biology of the Cell*) showed that increased concentrations of MSUD metabolites, in particular a-keto isocaproic acid (KICA), specifically induced apoptosis in glial and neuronal cells in culture. Apoptosis was associated with a reduction in cell respiration and, significantly, KICA also triggered neuronal apoptosis *in vivo* following intracerebral injection into the developing rat brain. These findings suggest that in MSUD children, neurodegeneration may result, at least in part, from mitochondrial damage due to the accumulation of BCAA and BCKA.

In the future, we aim to focus on the precise mechanism of KICA toxicity. We have already ascertained that KICA-induced apoptosis is not accompanied by the release of cytochrome c from mitochondria to the cytosol, a common occurrence in apoptotic cells. This suggests that an alternative pathway for apoptotic execution exists. Our goal is to dissect this death pathway in vulnerable brain cells exposed to physiological combinations of BCAA and BCKA.

With this approach, we hope to identify appropriate therapeutic targets to inhibit or delay apoptosis. If this strategy succeeds, there are potential implications for cerebroprotection in

MSUD children. The brains of babies are less than one fifth the size of adults'. Saving a few thousand brain cells in MSUD babies from apoptotic death would result in millions of cells in the adult that would otherwise have been wiped out by the accumulation of BCAA /BCKA. In this way, the neurological deficit associated with MSUD would be minimized.

Our findings may also have implications for disease management. Diet is critical in MSUD patients and the vast majority of patients manage the disease successfully by carefully restricting BCAA intake. Nevertheless, even with the most diligent care, children can sometimes undergo a metabolic crisis. These episodes can result in brain damage if undetected and untreated. Understandably, the constant threat can put immense strain on the families of MSUD children. If our preliminary findings are confirmed, anti apoptotic therapy may be a way to minimize cell damage resulting from BCAA/BCKA accumulation. We hope that the potential clinical applications of our research can offer these children and their families the chance of a better quality of life.

INTERNATIONAL NEWBORN SCREENING MEETING

Details

Written by Sandy Bulcher

Published: 20 July 2009

I have been interested in newborn screening since our son Jordan was diagnosed with MSUD 12 years ago. Ohio was not screening newborns for MSUD at that time. (Screening for MSUD in Ohio started in January 2001). I felt very angry after learning that screening was possible for MSUD, and Jordan suffered needlessly as a result of a delayed diagnosis.

Initially, I was only interested in advocating for MSUD screening. However, once I met children with other diseases who suffered as a result of the lack of screening, my vision expanded. I now know that it is essential for all babies to receive comprehensive screening to ensure their well being.

Several private labs do comprehensive screening - it is possible to screen for 30 or more disorders. Also, several state labs offer comprehensive screening to all the babies born in their states. Most state labs, however, continue to screen for 4 to 8 different diseases. MSUD is screened for in only 21 states.

The first national meeting regarding the use of tandem mass spectrometry (MS/MS) for newborn screening was held in San Antonio, Texas in June 2000. At that time, three states

(Massachusetts, North Carolina, and Wisconsin) and two private institutions (NeoGen Screening and Baylor Institute of Metabolic Diseases) were using MS/MS for newborn screening. Since June 2000, several state programs (Iowa, Minnesota, Ohio, and South Carolina) and one private institution (Mayo Clinic) have become operational. There are several states that are in various stages of adding MS/MS technology - California, Illinois, New Jersey, and New York. In addition, most babies in Pennsylvania receive comprehensive screening through NeoGen Screening.

In September 2001, I had the privilege of attending a newborn screening conference in Madison, Wisconsin titled, "Enhancing the Implementation of Tandem Mass Spectometry for Newborn Screening Laboratories." The meeting was hosted by the Wisconsin State Laboratory of Hygiene.

The purpose of the meeting was to share experiences with using MS/MS technology. Speakers shared their expertise on a number of issues including: implementation of MS/MS technology, follow-up plans, diagnosis, and treatment of the disorders identified by tandem mass spectrometry.

Many professionals involved with newborn screening from all over the country, as well as many international lab directors, were present. There were only three parents present - Trish Mullaley, PKU mom from Massachusetts representing the National Coalition for PKU and Allied Disorders, Tera Mize, from Georgia representing Saving Babies through Screening (formerly Tyler for Life), and myself representing the MSUD Family Support Group.

The first day (September 10) was packed with information. I had an opportunity to meet a number of state lab directors and others from the private sector. That evening, we toured the Wisconsin Newborn Screening Lab.

The following day (September 11), the meeting barely got started when we learned of the terrorist attack in NewYork City. As you can imagine, the tone in the room changed dramatically after the announcement. We were dismissed for an hour or two to digest the news. Like many of you, I was glued to the TV as I watched the events unfold. Suddenly, newborn screening, and most everything for that matter, seemed very unimportant. All I wanted to do was go home. We were told no planes were flying and no trains were available for transportation. I was fortunate, however, to find space in a rental car that was heading for Columbus. The majority of the meeting participants were from other states or countries, and many did not have transportation home. The meeting continued for those who were staying, but many participants left early as opportunities arose for transportation.

I really enjoyed the meeting in spite of missing most of the second day. I encourage all of you to question your state lab. Do they have MS/MS equipment? If so, where are they in the process of implementing MS/MS screening for newborns? To find out which disorders your state screens for, see: www.savebabies.org

PERSONALLY FROM THE BRUBACHERS

Details

Written by Wayne, Joyce & Shayla Brubacher

Published: 20 July 2009

Newborn Screening Experience
We were blessed with the birth of our 3rd grandson on Jan. 20, 2002. This gave us another opportunity to observe the process of newborn screening. If our grandson had been at high risk, we would have had him checked through the Clinic For Special Children in Pennsylvania at 24 hrs. of birth or sent a blood test immediately after birth to the lab in Columbia, Missouri for DNA testing. With either of these tests, results are available the next day - at two to three days of age. Instead, we made sure he was tested for 30 diseases by Neo Gen Screening in Pittsburgh, Pennsylvania as well as through our Indiana state screening program which includes MSUD.

One and one half years ago when our granddaughter was born, it took five days for the blood sample to reach the laboratory in Indianapolis. This time, with our grandson, blood was taken and sent to the state lab via mail from our local hospital two days after birth. The state received it *6 days* later. Both times the test results were available from the state lab the following day. They assured me that a positive result would be faxed immediately to the doctor. However, since both grandchildren's tests were negative, they were mailed to the local hospital where *the doctor picks them up when he makes his rounds* and the results are then reported to the parents. Our son and his wife received our granddaughters report two weeks after birth and our grandson's lab report four weeks after birth. The most important is the return time for a positive test - possibly *the earliest* for our grandson would have been at 8 days of age. Is this acceptable?

Blood was taken from our newborn grandson for the Neo Gen screening test on the third day. (The instructions say the "ideal time to take the blood is between 24-48 hours of age, as close to 48 hours as possible.") We sent the sample via FedEx the same day it was taken. Neo Gen received it the following day, and the results were available the 5th day. However, the instructions say, "Abnormal results are available 3 working days after the receipt of the sample and are called directly to your physician. Normal results are mailed to your physician. It may take 2 weeks for them to receive the mailed results." If we had sent the specimens via mail as instructed by Neo Gen, the results would have been delayed several days.

Neo Gen has 3 testing options. (1)The MS/MS (45 disorders for $24.50); (2) Supplemental packages only available for children less than 7 days of age (55 disorders for $49.50); (3) High risk screening for older or ill children (65 disorders for $59.50). [Check their web site: www.neogenscreening.com or e-mail: info@neogenscreening.com.]

Recently, near Philadelphia, parents were notified when their son was 10 days of age that he had MSUD. This child was detected through the Pennsylvania state screening program. This time frame seems to be typical of current screening programs. Although certainly better than no screening, this turn around time issue needs to be addressed. Is the big issue the cost of overnighting specimens? How does that cost compare with the risk of brain damage caused by several days of delay?

NEWS & NOTES - BEHAVIORAL STUDIES

Details

Written by Joyce Brubacher

Published: 20 July 2009

When parents of children with MSUD notice puzzling, abnormal behavior, it raises many questions. Is the behavior related to their metabolic condition? Do other parents notice the same behaviors? Do all children with MSUD manifest the same behavioral patterns or do the genetic mutation, the amount of damage before diagnosis, degree of metabolic control, protein deficiencies or other factors make a difference?

Hopefully, a proposed behavioral study will provide some insight on MSUD behavioral issues. Dr. Dean Danner from Emory University in Atlanta, Georgia and Wendy Packman, J.D., Ph.D. from San Francisco, California plan to do the study with the cooperation of the MSUD Family Support Group. The Support Group has given some initial financial support to begin the study. The doctors will soon send questionnaires to validate the results of an earlier study begun in 1996. No one is obligated to participate, but a quick and thorough response would be very helpful. Dr. Danner will report on the progress of the study at Symposium 2002 in July.

The study will use standardized child assessment evaluation forms filled out by parents and/or the affected individuals along with a companion form filled out by the child's teacher to evaluate behavior patterns. The completed forms will be scored and evaluated independently by a Certified School Psychologist and a Clinical Psychologist.

The results of this study will provide the first evaluation of behavior patterns in these individuals. Their molecular genetic abnormality has already been defined by completed laboratory studies and the results of the behavior study will be aligned with their specific gene defect. The benefit will be felt most readily by new individuals identified with this genetic disorder. In addition, caregivers will be able to anticipate and therefore better manage the psychological needs of these individuals.

NEWS & NOTES - NOTICE OF CHANGES

Details

Written by Joyce Brubacher

Published: 20 July 2009

Notice some changes in the list of contact persons on the cover of this issue of the Newsletter. Sandy Bulcher is now listed as a contact person. She is very knowledgeable about newborn screening issues as well as a resource for any MSUD-related information for families and professionals. Her husband, Dave is our treasurer. They are accessible by phone, e-mail or regular mail.

Dawn Hahn is still available as a contact person, especially for families in Pennsylvania for family information packets. Her address will be changing in the near future.

The area code for Goshen, Indiana has changed. Be sure to change this on all contact information for Wayne and Joyce Brubacher. The area code is now 574. (However, the old 219 can be used until June.)

NEWS & NOTES - MELISSA & JESSICA

Details

Written by Joyce Brubacher

Published: 20 July 2009

Art Award For Melissa
Melissa Berman, Peabody, Massachusetts, daughter of Bob Berman was recognized for her art talent in the Sixth Congressional District Art Awards. A student at Peabody Veterans Memorial High School, she was named "best of school winner." Melissa, 20, has classic MSUD. She sold a painting of hers in December for the first time, and she received $60 for it. Melissa intends to pursue a career in art. We extend a hearty congratulations to her for her accomplishments.

Jessica Featured in *Reaching Out*

Jessica Berman, 17, is Melissa's younger sister and also has MSUD. Bonnie Brien is Jessica's "Big Sister." Bonnie has been including Melissa in their adventures and even accompanied them to Symposium '98 in Lancaster, Pennsylvania. The girls' father Bob is thrilled that Bonnie is a "Big Sister" to Jessica and is pleased Melissa is included in many of their outings. (Melissa and Jessica's mother died soon after Jessica was born.) Jessica is on the left in the photo above and Melissa on the right. Bonnie is in the middle - a good friend to both girls.

Jessica was named Student of the Month at her school in October 2001. Our sincere congratulations to Jessica. Keep up the good work.

SHARING - WHEN THE STUDENT BECOMES THE TEACHER

Details

Written by Susan Jasin

Published: 20 July 2009

As we all are well aware, having children such as ours requires a lifetime of attention-to-detail and a never-ending commitment to learn. For most of us, our children's lives begin with us in shock and in a perpetual state of "what if I do it wrong"? We look to our doctors and the support group to answer all of the thousands of questions that arise from having a child with special needs. As time passes, and our children age, we develop a greater understanding of the care they require after experiencing illness, growth spurts, and the simply unexplainable fluctuations of leucine levels. In short, we the parents become the knowledge brokers to help facilitate the doctors and nutritionists ability to heal our children.

Like any parent, I am involved in all aspects of my son Jakob's life, and I am always searching for new information on MSUD, genetics or anything applicable to his disease. I have developed a close working partnership with his nutritionist, Kathy Camp, at Walter Reed Army Medical Center in Washington DC. In light of our relationship, Kathy asked if I would be interested in presenting Jakob's case, "a snapshot in the day of," sort of thing, to a group of Dietetic Specialists at the American Dietetic Association Convention in St. Louis, Missouri, October 20, 2001. Since my husband and I are originally from St. Louis, and it was a chance to educate others as well, I put aside my fear of speaking in front of large groups and agreed. The specific class that I would be speaking to was titled "Applying Genetic Principles to Dietetic Practice."

My portion of the class was twenty minutes - focusing on my perspective of MSUD and how dietitians can, and have been, helpful to my family and me. With this as my guideline, I chose to concentrate on the importance of the Nutritionist in helping to manage Jakob's disease. (Have I said before that I am not comfortable speaking in front of groups of people?) I was very nervous in the days prior to the conference and rehearsed ahead of time what I wanted to say. My husband kept telling me that I could talk for hours on end, in intricate detail, about Jakob and MSUD. He said the most difficult part would be getting my point across in only twenty minutes. Still, nothing like being prepared. I practiced and practiced, I brought along pictures and I discussed the "tools of the trade:" formula, mixing equipment and scales. I also came armed with a handful of Newsletters and brochures courtesy of Joyce Brubacher!

Beginning to speak, it was hard to fight back the tears as I outlined the diagnosing process we went through with Jake. I realized that after two years I am still healing from that experience, and I have not fully worked through all of those issues. Next I spoke of the daily stress that we as parents live under and how much trust we place in the "professionals" that treat our children. More than anything, I wanted to convey to the attending nutritionists and dietitians the need to listen to the parents and for them to treat the patient, not just the disease. I wanted them to know that each child with MSUD is different and has their own specific needs from treatment. We as parents watch our children grow and observe the slightest change in behavior or even the onset of an illness better than anyone else. We are the advocates for our children and we must be included in their treatment.

It is daunting to show a group of educated, medical professionals that "thinking outside of the box," by including parents with little or no formal medical training, is really in the best interest of the children. I know that I was successful due to the number of conference participants who approached me when the lecture was over. To a person, they thanked me for speaking, they asked more in-depth questions, and some of them were as misty-eyed as I had been when I first began to speak. Knowing that I was able to reach people and help them understand a complex problem, and knowing that they in turn will use that knowledge to the benefit of others with MSUD, helps me in my own healing process.

Jakob will be two years old on the 27th of January and my husband and I have learned much in that time. Like many of you reading this, we feel frustrated at times with family and friends who "just don't seem to get it" when it comes to MSUD. Combine that with the misinformation that often gets reported in the news,* and we realize that we all need to be advocates. Each doctor, nutritionist or dietitian we make smarter is one less child that will be at risk because of MSUD. I may not get the opportunity again to speak in front of such a

large audience about Jakob or MSUD. However, I will continue to learn what I can and I will share that knowledge with whoever should seek it.

**An article in the St. Louis Post Dispatch, (1/6/02) reported on Dr. Hillman's metabolic clinic in Columbia, Missouri and the simple "cheek swab" DNA testing done in his laboratory to detect the Mennonite mutation of MSUD. Some information in the article was inaccurate and misleading. It did a great disservice to the Clinic at the University of Missouri.*
- Editor

SHARING - FLU SHOTS

Details

Written by Kay Larsen

Published: 20 July 2009

An issue discussed in the MSUD eGroup was immunization for influenza (flu) each fall for children with MSUD. Kay Larsen shares information that has helped her as the mother of Maritsa, 24, who has classic MSUD. Maritsa is adopted and has neurological damage due to her early history. Maritsa has thrived under Kay's vigilant care.

I always make sure that my daughter gets an influenza shot before winter - usually in October or November. It takes three to four weeks to build up antibodies after the shot is given. The shot should be given well before flu season actually starts.

I appreciated an article in a newsletter from the metabolism department at Children's Hospital of Philadelphia, Pennsylvania.

It was written by Alice Mazur, RN, MS, CRNP, who was working in the department of metabolism at that time (1996). The article reads:

"Flu season is also coming. We urge you to have your child receive this year's flu shot. Each year predictions about which flu virus will be most prevalent are made by experts at the Center for Disease Control. They provide that information to the pharmaceutical companies, who then produce and distribute them. We believe that your child should be protected from the flu, so that severe metabolic decompensation does not occur. Each year, some children become ill from the flu and require hospitalization. We would like to prevent your child from becoming ill and the flu vaccine is a good way of preventing illness. Contact your local pediatrician, family doctor or local health department about scheduling the flu vaccine. Most health insurance carriers cover the cost of the shot. Some children do not respond well to the flu vaccine. If your child has gotten slightly ill after the flu vaccine has been given in the past, there are some steps you should take.

Decrease the protein in the diet on the day of the flu vaccine. One-half of the total protein is usually sufficient.

Give actetaminophin for fever and/or discomfort. This may be given before the shot and then given every four hours after the vaccination during that day.

Give your child extra fluids and calories on the day of the vaccination. It is usually best to use the special metabolic product (formula). If your child is not receiving a metabolic product, use high calorie, low protein drinks and foods.

Call the metabolism division if you have any questions or concerns."

I have used this advice for my daughter every year and she has not had any problems at all when receiving the shot. In addition, her regular doctor started out by dividing the vaccine into two doses instead of one - with a week or two between doses, just in case she should have a bad reaction. This requires two injections and two trips to the doctor, but is well worth it if you are nervous about giving the vaccine as I was initially.
The bad reactions that Alice talked about are usually (but not always) mild ones, consisting of soreness and redness at the injection site and mild fever.

Fortunately, either because of the vaccine or by God's grace or both, my daughter has never had the flu. And she has never had any bad reaction to the vaccine, probably because I follow these recommendations carefully.

Hope this information is helpful in some way. I would definitely urge you to show these recommendations to your child's doctor before you try them.

FEATURED FAMILIES - JOSHUA HILDYARD - AUSTRALIA

Details

Written by Susan Carter and Darren Hildyard

Published: 20 July 2009

The day Joshua was born was a very happy one, June 23, 2001. He was a healthy little boy and took to the breast straight away. All went well with feeding and everything seemed perfect. After about one week, Joshua fussed at the breast and seemed to have forgotten how to feed. We were becoming concerned about his feeding, and our baby Josh was losing weight. He still hadn't regained his birth weight. Josh was put on the bottle as breast feeding didn't seem to be satisfying him - at least we could see what he was actually

getting. Josh started to scream all the time. He would sleep for about 10 minutes to about half an hour and then wake up. His body would go stiff when he cried, and his arms moved as if he were climbing a rope. We thought he had really bad colic. The midwife was becoming concerned and helped us as much as she could. We even took him to our doctor, and he seemed to think Josh was fine.

At two weeks of age, we received a phone call from a doctor from Elizabeth Hospital where I gave birth to Josh. He asked me to come to see him straight away. Once there, I was informed that Joshua's Guthrie test had shown he had MSUD. Thankfully, South Australia has offered tandem mass spec acylcaruitine and amino acid profile newborn screening for the last two years.

This news was devastating: this beautiful little boy had a disease for the rest of his life. Why Josh? We were then referred to the Women's and Children's Hospital where we met Doctors Janice Fletcher and Michael Metz from the metabolic unit. Treatment started straight away and slowly, but surely, Josh changed from a screaming, irritable baby to a very contented baby.

The Metabolic team is fantastic; just seeing their faces when Josh is sick makes us feel so much better. We still have a lot to learn about MSUD, and some of the things we have already learned are pretty scary, but Josh has handled his first virus and immunization very well.

At first we didn't like taking blood from Josh, but now it doesn't bother him and is part of our regular Sunday night routine. Now we just remind ourselves that this is a necessary part of Joshua's life. As Josh is only one of two people in South Australia with MSUD, there is little experience and only limited information on MSUD here. So we surfed the Internet searching for more. Thankfully we found the MSUD Family Support Group site. After reading all the newsletter articles on the site, we now have a better understanding of MSUD and what to expect in the future. Although not all the news was good, we know that it's not all bad either, and we are better prepared for the days ahead.

We were very happy to meet Kym and his family recently. Kym is 15 and the only other MSUD case in South Australia. It was reassuring to see such a healthy, energetic young man with the same problem as our Josh, and to see he is doing so well.
Josh is now 10 weeks old and doing very well. He loves his bottle and is putting on weight every day and responding to us with big, beaming smiles. His progress is that of a normal, healthy, little boy with no apparent damage from the early effects of MSUD.

We are very thankful that Josh has such a wonderful group of doctors looking after him - not only that, but they look after us, too.

FEATURED FAMILIES - NOAH WEBER - ONTARIO, CANADA

Details

Written by Naomi Weber

Published: 20 July 2009

Noah is the son of George and Naomi Weber. Naomi has shared the story of their growing family in the Newsletter through the years. Daughter Lydia wrote a candid account of living with MSUD as a teenager for the fall/winter 1997-8 issue. Lydia (18) bakes low protein items for Noah (8), brother Benjamin (21), and herself - the three who have MSUD in their family of 10 children. (An older brother, with MSUD, died in 1982 at the age of 4.) The family is Orthodox Mennonite; they use a horse and buggy for transportation, hence Naomi's references to hiring drivers to take them a distance. Naomi refers to her husband George as Dad throughout this article.

In the early morning on November 16, 1993, our eleventh baby, Noah, was born. He was a chubby little boy who seemed very quiet and relaxed as he lay at my side. The first and second days slipped by. We kept a close eye on him as we didn't send a blood sample to be tested as we usually don't get results very soon anyway.

At fifty hours, I started to wonder as I noticed the baby looked upward and also bit me when he fell asleep while nursing. The next several hours, he still seemed quiet, and we still hoped. . . .

Then at bedtime that evening, Noah got his first high-pitched cry, arched his back, and we couldn't quiet him right away. I then mixed a high calorie drink and gastrolyte solution. He took a few ounces every few hours during the night and had no more crying spells. As we didn't have valine and isoleucine on hand, and no BCAA were mixed with his medical food, his valine and isoleucine went flat, and the leucine was sky high. He soon developed a bad rash around his mouth. We thought we could possibly send a blood sample on the bus going into Toronto.

So early the next morning (11/18), we took the sample and packaged it with some ice in a cup-size thermos. Dad drove the ten miles to catch that bus. At 4 p.m. the same day, we got the report. Noah's levels were up to 975 µmol/l already. By day six Noah was too drowsy to take all the formula. The blood test done that morning was 1480 µmol/l. I asked for valine and isoleucine, but it would take 24 hours to get it. We needed the supplements sooner. I realized we were losing out now. It was late evening, so we would wait till early morning to leave for the hospital in Toronto. We asked my brother Emanuel to put in an NG tube for the

night. (He learned to insert an NG tube in his daughter with MSUD.) He and his wife Lovina fed Noah gastrolyte solution every two hours all night.

We arrived at the Toronto hospital at 7:30 a.m. the next morning. Noah had slept since 3 p.m. the day before, and he had a level of 1116 µmol/l. Valine and isoleucine were added to the gastrolyte solution in the NG pump. After 24 hours, the levels dropped to 875 µmol/l. The formula was no problem then, and Noah was our contented baby again.

Noah had only a few sick days, but nothing really serious until in August 1998 when he had his second hospital stay. He had fever, vomited, then got so tense and restless; he cried and held his breath - a slight seizure. He was on IV for 24 hours and then was himself again. He actually thought it was fun in the hospital with nice toys, books that talk, and a bed with buttons to press to go up and down. We felt quite lucky that the IV treatment helped so well and that it was not a long stay.

In the winter of 1999-2000, we experienced a very hard flu in the community - people were in bed for a week or so. And then Noah got it. On January 23, 2000, Noah had a croupy cough with fever. The second day, the DNPH was still clear; he took mostly fluids and ate low protein Jello. Then in the evening, he started to vomit. The suppositories we used didn't settle him for long, and he vomited again.

On the 26th at noon, Noah lay more quietly and stared; he didn't seem to respond well when we talked to him. He refused his drinks. Again, the decision had to be made: go to the emergency room in Wingham hospital. The nurse put in an IV, but not as fast as needed, and not enough calories. We waited five long hours to finally see a doctor. A doctor who didn't realize he had a serious case. He contacted Toronto and got the recipe for the high calorie solution. Then we were admitted and put into a small room on the second floor for the night. Noah showed no more ketones, but he didn't sleep too relaxed. Neither did we - an old lady who talked all night was a roommate!

The next day, Noah seemed to be the same until 7:30 when he had a seizure. That really concerned me - and his fever was up. The doctor called Toronto; the doctors there still felt his fever could be causing the seizures. Noah slept fairly well, but woke early. He was frightened of falling and was seeing eyes everywhere. I called our neighbor to bring Dad and the DNPH solution. The test showed Noah's levels were very high. All along Noah had complained of abdominal pain, which was also a concern.

Arrangements were made to transfer Noah to Toronto. His dad went along, and I went home to get some rest. In the evening, our son called to find that Noah was still having seizures and his levels were 1300 µmol/l . Besides regular IVs, he was given lipids and TPN. The next morning, Noah had improved only a little.

Friday, I went down to the hospital. Noah looked happier again, but still complained of pains in his tummy. He often asked to have it rubbed. Saturday, he was out of bed and started eating and drinking. The nights were wearisome with Noah jumping and crying as though frightened by a bad dream. I slept in bed with him to hold him down.

Sunday, Noah woke up sick again with high fever. More x-rays of his abdomen. His appendix was enlarged and dislocated. By evening, the doctors had made plans to remove

the appendix Monday morning. Seemed our cloud wasn't past yet. The poor suffering boy, why the surgery? And Dad cannot come as he is down with the flu.

On Monday morning, Noah was much better and his levels were down to 600 µmol/l - and I thought we could go home! As Noah ran down the hall for his bath, I thought he must go to surgery in an hour. But Noah wasn't hesitant. He wanted the doctors to do something about his pain.

Noah bravely went with the OR nurse. My sister-in-law and I were taken to another room to wait for the doctor's report until we could see Noah in recovery. My brother came in Dad's place to be with Noah the next day. I had to leave Noah, weak, helpless, and sore.

In the following days, Noah improved steadily and was ready to go home on Friday. When the surgeon discharged Noah, he said he might never see us anymore, and I wished we wouldn't have to see him either! But as the year rolled around and March came, we found ourselves back in the same room and bed, and had taken the same route as last year.

Noah entered Wingham hospital with severe tummy pains and vomiting. After 48 hours of IVs and many calls to Toronto, we headed down to the Toronto hospital again.

Noah was vomiting green and black stuff; tubes were pushed into his stomach to draw the mess out. By Saturday evening, Noah started to seizure, then finally he was taken to surgery. They found scar tissue grown around the bowel obstructing ten inches which had to be removed. Noah's levels were up to 1100 µmol/l, so to be on the cautious side, he was put on the hemodialysis machine.

Early Sunday morning, our kind neighbor drove my brother and sister-in-law and me down to Toronto to see how Noah was and to stay with him for a few days. We found him in the Critical Care Unit, hooked up to all kinds of machines and IVs. He was taking three different antibiotics. The dialysis needed was fixed in his chest. He was drowsy yet from the anesthesia; when he was awake, his mind was in confusion from the morphine he was getting. He thought he was so hungry and thirsty. Of course, his stomach felt miserably empty with the suction tube in. I dampened his lips with ice water on a little sponge which he would almost eat. We tried to keep all food and drink out of his sight. (By Sunday evening, Noah's levels had come down, so he was moved back up to the genetic ward.)

Tuesday evening, I went home. Then Dad stayed with Noah until Friday when I came to be with him again. By Friday, Noah was taking small walks. We hoped he could start taking formula, but it made him sick to his stomach, which showed he wasn't ready for food.

On Saturday, the dialysis tube, through which Noah was getting lipids and TPN, fell to the floor when we were walking. The nurses got quite excited, grabbed him up and put him back to bed. The doctor was called. The bleeding soon stopped, but Noah's feedings were off. He was taken to surgery to put in a different line - one that would take thicker solutions. The doctor felt he wouldn't need to anesthetize Noah if I would keep him quiet in the operating room. The doctor said it wouldn't take long, but Noah was very worried and upset. He needed some coaxing and stories to finally relax as he was strapped to the table and covered up. And I was dressed like a surgeon myself! Without being drugged, it sure didn't take long to put in a central line. The needle was now in his arm instead of the chest.

Noah seemed to improve after the second weekend. I felt I should stay to give him his feedings and meals. The hospital cook was not trained to prepare the special low protein foods, although they did mix Noah's formula. As it was, by the time Noah could eat, his levels were down so much, he could go right on his regular diet - Rice Krispies, potatoes, carrots, etc. How Noah enjoyed eating again after one and a half weeks with no food!

Wednesday, two weeks after being admitted to the Toronto hospital, we had high hopes of getting to go home. The surgeon was pleased with Noah's progress and discharged him. But the central line was still in Noah's vein. We waited to have it pulled out.

Our neighbor had brought Dad and our three school-age children down to the hospital to take us all home. We packed up all our belongings, Noah's teddies, and even a quilt. But to our great disappointment, when five o'clock finally came, the surgeon refused to pull the line out as Noah had been on solids only two days - he wanted to be sure.

Now what shall we do? I was so tired and listless from sitting around, I couldn't even cry. It was decided that Dad and I would stay with Noah. The children bravely went back home with our driver [their neighbor]. They were met by the rest of the family long waiting for us all!

Such is hospital life with so many doctors involved, but we were deeply thankful Noah was this far recovered, and we could easily praise God even in our disappointment. The next day at the hospital went better, and we went home on the evening bus.

We continued with weekly blood tests for a few months as Noah's leucine levels didn't go above 100 µmol/l even with 14 grams of protein a day. Noah's body was using a lot more protein to repair the wounds and build up the muscles which he lost lying in bed almost two weeks. Noah's hair got really thin, and he was afraid of becoming a baldy! Just three months after surgery, his levels were finally where the doctor recommended.

Noah enjoyed going to school after his long leave. He was promoted to grade two in June [2001]. I think his teacher sometimes finds it difficult as Noah gets distracted easily and can be so restless - he is in a class with four other little boys. We can only be thankful that Noah is able to run and has the energy after all he went through the past two years. What is ahead we do not need to know, if only we can in patience teach him and love him for the Lord...

The treatment described in this article is not necessarily standard treatment. Personal choices and hospital protocol do vary.