VOLUME 10-2

Fall/Winter, December 1992

This issue includes a brief review in words and pictures of Symposium '92. Dr. Elsas shares some exciting news on his latest research project and Dr. Morton describes the medical implications of brain surgery on a young boy with MSUD.

FAMILY HISTORY - JORDAN GROFF

Details

Written by Jordan Groff

Published: 14 July 2009

The following history was written by Glenda Groff from Pennsylvania, mother of Jordan. She enclosed a photograph of Jordan sitting on the floor with a box of spilled cereal. He has a very mischievous look.

On June 29, 1991, our son Jordan was born. He seemed to be a healthy child. We returned home from the hospital the next day. Jordan received the newborn screening test at 24 hours just before we left.

He never nursed as well as our daughter had. On July 4th I first noticed his breathing seemed really irregular. The following day the health care nurse came to our home. She stated that she thought everything seemed fine. Being reassured, I decided I must just have a fussy baby.

Saturday morning we received a call from our family physician, asking a few questions. Little did I know the answers I gave told him what he needed to know.

Jordan was the first child picked up through the Pennsylvania state screening program. He was admitted to the Children's Hospital of Philadelphia. There we began to learn about these new problems we had to cope with. His highest level was 34 mg/dl Saturday evening. We brought him home 10 days later with a level below one.

We now had a very content baby who could hardly get enough to drink. All went well except for a few bouts of flu.

In March he got an ear infection. This was one of many, we were soon to find out. We were constantly going to the doctor and changing medicine, with good results for a short time,

then back again. Doctor Morton gave him several antibiotic shots, and they would carry him over a few days. We were thankful his levels did not elevate during this time.

On September 8th we went to see an ear specialist. We returned the following day and had tubes put in his ears. He came through the surgery really well. I gave him the same amount of formula he had been taking. His level remained at 1 mg/dl. Since than he is doing very well and has not been sick

Developmentally he is doing great. He began walking at 11 months and it's been a real chase ever since. His favorite thing is climbing, the higher the better.

Jordan is doing very well at this time with his diet. We have graduated him to a sipper cup. But it seems to be just Mom who he will take it from. French fries, squash, and applesauce seem to be his favorite foods.

We have learned a great deal about this disease. Jordan has the classic MSUD. It really is amazing to me how little people know about MSUD. In the Surgical Center, where the tubes were put in his ears, I had to describe this disease to everyone. Sometimes the reaction you get is very interesting. This remark came to me from someone, 'Oh, you mean he can't have maple syrup?' So we are continuing to learn more about MSUD.

We are thankful he is doing so well and for the insight he gives us into what other families go through with their special children. We also are thankful for our 3-year-old daughter, Andrea, who doesn't have this disease. A special thanks to Dr. Morton and his staff and all the support we get from the families.
May God bless each one of you, Ernie, Glenda, Andrea, and Jordan Groff

MSUD NEWS FROM THE CLINIC FOR SPECIAL CHILDREN

Details

Written by Holmes Morton M.D.

Published: 14 July 2009

Amos Fox helped chore as usual on the family farm this summer. It could have been quite different for him and his family if it hadn't been for Dr. Holmes Morton. This article is an unedited account of Amos' successful brain surgery as written by Dr. Morton for this Newsletter. Hopefully what was learned from this experience will be of value in treating other children with MSUD. Thank you Dr. Morton for taking time to share this important information with us.

Last Spring I was asked by Joyce, Alma and Weaver Fox, and others interested in the case to write about the hospital management of a 13 year old boy with classical MSUD who underwent surgery to remove a brain tumor. I have found it difficult to write a summary of the case because details of his treatment were complex and my conclusions preliminary. Nevertheless, I think the case raises important questions about treatment of MSUD which parents, patients, and physicians who read the MSUD Newsletter should be aware.

DAILY MONITORING OF MSUD AND RECOGNITION OF UNDERLYING ILLNESSES:
The earliest sign of Amos' tumor was difficulty with control of his MSUD. Alma found that the urine DNPH test was positive regardless of changes in diet which previously brought his MSUD under control. If Alma had not used DNPH tests to monitor Amos, and had she not made repeated efforts to improve his metabolic control, then the diagnosis of the tumor would certainly have been delayed. As discussed below, the pressure of the tumor upon the brain stem caused vomiting and such severe metabolic derangements that without Alma's careful monitoring I think Amos might have died from metabolic intoxication before the underlying cause was discovered.

Although Amos' tumor was an unusual cause of loss of metabolic control, his case is an example of the importance of daily monitoring with DNPH and careful dietary records for the general health care of these patients. When a patient who has been under good control with a fixed leucine intake of 20 mg/kg-day becomes leucine intolerant as shown by repeatedly positive DNPH tests then their management must immediately be changed and a reason sought for the loss of metabolic control. More common causes of sudden changes in the leucine tolerance are painful injuries, viral infections such as influenza, childhood hepatitis, immunization reactions, hidden dental abscesses, chronic inner ear, sinus or urinary tract infections. Uncommon but by no means rare problems such as hyperthyroidism and insulin dependent diabetes would also cause loss of metabolic control in MSUD patients long before other signs of these disorders would lead to diagnosis. Monthly or twice yearly blood leucine measurements are not adequate for early detection of such problems. I have no doubt that careful control (and records) of daily leucine and caloric intake and routine monitoring of urine DNPH will decrease the frequency of life-threatening metabolic illness in children with MSUD and will help early recognition of important underlying conditions.

LIFE-THREATENING METABOLIC ILLNESSES PROVOKED BY SURGERY OR INJURY:
Surgery or injuries that involve prolonged, stressful, or painful recoveries are especially dangerous to children who have MSUD. Plans to monitor and manage MSUD must be made carefully. Throughout Amos' hospitalization urine DNPH tests and serum amino acid analysis were done 1-4 times daily as needed to monitor the frequent changes in therapy. Serum branched chain keto-acids were also measured to study the relationship between the urine DNPH reaction, blood amino acids, and keto-acids. MSUD hyperalimentation was started through a secure central venous line before surgery. His surgery was delayed until the serum leucine was 5 mg/dl. Even before his operation Amos' leucine level was difficult to control because of the tumor and because of the effects of a steroid called Dexamethasone which was given to reduce the brain swelling caused by the tumor. Although this steroid is known to stimulate the breakdown of protein, Amos' neurosurgeon,

Dr. Edward Garitto, felt the medication was necessary, and I agreed that with careful management of MSUD the likely benefit of the medication outweighed its risk.

The brain surgery and the first 12 hours after surgery passed without difficulty. The earliest sign of worsening metabolic control was increased blood glucose and resistance to the insulin which was added to keep the blood glucose in the normal range. Over the period 12-24 hours after surgery the DNPH test changed from clear to cloudy and leucine increased from 6 mg/dl to 10 mg/dl regardless of MSUD hyperalimentation and high doses of insulin. Although it was possible to prevent further increases in leucine, I was aware that the nutritional support necessary to control his MSUD was far greater than expected. In other MSUD patients, in and out of hospital, I have found that caloric intakes of 1.5-2 times the calculated basal metabolic rate of the patient will stop protein breakdown and usually steadily lower serum leucine. Amos' basal metabolic rate when he is well is approximately 28 Cal/kg per day (1000 Cal/day). My initial goals were 60 Cal/kg-day as glucose and 1 gram/kg-day protein in the form of a MSUD amino acid mixture for intravenous use. By day #3 after surgery serum leucine level could just be kept below 12 mg/dl with hyperalimentation rates of 110 Cal/kg-day and 1.8 g/kg-day of MSUD amino acids combined with high insulin infusion rates (0.5 units/kg-hour). Dexamethasone was stopped 3 days after surgery because of my concern about the difficulty of lowering leucine. Insulin requirements decreased rapidly over the 24 hours after Dexamethasone was stopped, but the leucine remained very resistant to change. In 24 hours leucine could only be lowered from 10 to 8 mg/dl regardless of a MSUD hyperalimentation rate of 122 Cal/kg-24 hours and MSUD amino acid mixtures of 1.8 g/kg-24 hrs.

Although it was increasingly apparent that Amos was in an unusual high catabolic state, I questioned if the amino acid mixture in the hyperalimentation was for some reason ineffective÷in retrospect this was not true. To test this idea, on the 6th post-operative day I asked Amos (who was surprisingly cooperative) to drink 60 oz of MSUD formula (40 Cal/kg-day) and I provided another 35 Cal/kg-day as intravenous glucose. Despite this Caloric intake of 2700 Calories (2.5 times his normal basal metabolic rate) and the amino acid mixture in MSUD formula, his leucine increased from 8 mg/dl at 10 AM to 18 mg/dl at 4 AM the next morning and to 31 mg/dl 14 hours later. The DNPH test became strongly positive, and he became disoriented and showed other signs of intoxication. This was an extremely rapid and dangerous increase in leucine. Hyperalimentation was restarted with glucose infusion rate of 150 Cal/kg-24 hours, 4 gm/kg-24 hours of the MSUD amino acid mixture, insulin was used as needed to keep the blood sugar normal, and infusion of intravenous fat was started. The total daily caloric was more than 180 Cal/kg-day, nonetheless, his leucine level was again very difficult to control. Serum leucine decreased only from 31 mg/dl to 28 mg/dl between day 7 and 8 and Amos continued to show signs of intoxication.

It was apparent that Amos was in an extreme protein catabolic state which could not be controlled by the calories, glucose, amino acids in the hyperalimentation solution alone. The cause and the means to control the abnormal state had to be found. I knew that the effects of Dexamethsone upon glucose and insulin had been gone for several days, blood cultures were negative, there were no signs of infection in the surgical wound, and even his thyroid function tests had been checked and were normal. I reasoned that the abnormal hypermetabolic state must be related to the effects of surgery upon the brain-stem itself and most likely were caused by adrenalin-like chemicals released as a result of brain stem irritation or swelling. Hyperalimentation, intralipid, and insulin were continued and a

medication called Propranolol was given at the same dose used to control the severe catabolic effects caused by extreme hyperthyroidism. Propranolol at this high dose blocks the effects of the catacholamines which are adrenalin-like compounds released by nerves that originate in the brain stem and by the adrenal gland.

Within six hours after intravenous propranolol was given the leucine level had fallen from 28 mg/dl to 21 mg/dl and continued to fall rapidly÷serum leucine decreased from 28 to 12 mg/dl in the first 24 hours and 12 to 3 mg/dl in the next 24 hours. The urine DNPH test became negative when serum leucine was 15-12 mg/dl and isoleucine was 3.5-2.5 mg/dl. Propranolol was initially given intravenously and then was continued as an oral medication. Intralipid was stopped and hyperalimentation was slowly weaned over three days while MSUD formula and food were started. His DNPH remained clear and leucine levels ranged from 1-3 mg/dl until discharge with a daily calorie intake of 60-80 Cal/kg-24 hours, total protein 0.6 g/kg-24 hours and leucine at 10-20 mg/kg-24 hours. Amos remained on a well controlled diet and propranolol by mouth throughout his subse- quent radiation therapy. His DNPH remained clear and his average serum leucine level over this 3 month period was less than 4 mg/dl. Now 10 months later he is well, active, and there are no signs of recurrence of the tumor.

FINAL COMMENTS:

Physicians who work in neurological intensive care units are aware that patients often have severe protein wastage after brain surgery or injury. I think Amos' severe metabolic illness after surgery was similar. I would also speculate that a similar high catabolic state develops in the final stages of fatal illnesses of MSUD. As brain intoxication worsens and brain swelling develops, that catacholamines are released from the brain which stimulate extremely high rates of protein breakdown and rapidly fatal intoxica- tion. I expect that beyond a certain stage of illness the metabolic effects of these signals cannot be easily reversed by MSUD hyperalimentation alone as was seen in Amos' case. Control of such hypercatabolic states will require a combination of careful nutritional management and use of medications that block catabolic signals and stimulate protein synthesis. Insulin and propranolol were used in Amos' management and are inexpensive, relatively safe, widely available. Other medications such as growth hormone or more selective catecholamine or cortisol antagonists may ultimately prove even more effective.

The rate at which Amos' leucine fell between days 9 and 11 was extremely fast÷on day 9 the rate was 16 mg/24 hours, over a 48 hour period the leucine fell from 28 to 3 mg/dl or 12.5 mg/dl per 24 hours. The paper by Dr. Berry about MSUD hyperalimentation reported a rate of decrease of leucine of only 4.5 and 6.5 mg/dl per day. In my opinion this combination of MSUD hyperalimentation and medication to block catabolic signals will also prove to be more effective and safer than peritoneal dialysis. The rate of fall of leucine in Dr. Scriver's 8 cases treated with combined intravenous and nutritional therapy and peritoneal dialysis averaged only 4.5 mg/dl - 24 hours with a range of 1 to 10 mg/dl. I do not think that Amos' case could have been managed with peritoneal dialysis. As MSUD hyperalimentation becomes more widely available, I believe peritoneal dialysis will be considered an obsolete method of treatment for MSUD.

I have recently treated another severely intoxicated 6 year old MSUD patient with methods similar to those used in Amos' case with similar metabolic results and full recovery.

Although encouraged by the results, I must emphasize that my observations discussed here are preliminary and unpublished. Other clinical studies will have to be completed before the approach is accepted as standard therapy.

POST CONFERENCE FOLLOW-UP

Details

Written by Toths and Sullivans

Published: 14 July 2009

Anna Toth wrote a letter to the Newsletter families on Oct. 15, telling about the birth of the baby she was expecting in June, and she included other information from the Toths and Sullivans who hosted the Symposium.

Time does fly. My new baby is now four months old and I can't believe the conference has come and gone. Daniel Robin Nicholas Toth was born on Father's Day, June 21, 1992; a week late, but better than having been a week early at the conference. He is a beautiful, healthy boy and has given all of us great joy. Thank you for all of your good wishes at the Symposium.It meant a great deal to our whole family. I remember when I had Michael, our MSUD child, I thought my chance of having a full large family was so limited; only to look around now and, at times, with sheer disbelief realize all my dreams have come true.

We, the Toths, as well as Mike and Karen Sullivan, feel that the conference was a real success. The guest speakers were wonderful. Our sincere thanks to those who attended and shared with us. We felt the professionals were informal, warm and very approachable. Many of you expressed the feeling that meeting Dr. Menkes was a real bonus. Wasn't he just fascinating! It really impressed me that if it weren't for the curiosity of these dedicated doctors, this disease would not be as treatable as it is today. Dr. Elsas was pleased to have a gathering of children with MSUD and their siblings to further his study. I'm sure all who participated are eagerly awaiting the results. He and Dr. Klein were interesting and informative.

We were glad the meeting, meals and lodging were all in the same building. It made it much more convenient for all of us. We were so encouraged by the feedback that we received on your little yellow sheets of paper. It would appear that you all felt the speakers were very fun and interesting, and the food excellent with a very good variety for the MSUD children. We realize that without the daycare facility we provided, it would have been impossible for some of you to attend. With positive feedback like this, we feel all the effort and the fund raising was well worth it. It all came together with only a few difficulties.

To any of the families who had problems with reservations, etc., again our most sincere apologies. We tried our best to rectify any situations that came up. We realize the facility was small for our meals, but eventually we all ate and ran pretty much on schedule. You

were all very patient and kind.

As for the helpful comments for future conferences, we have reviewed them and passed the information along to those hosting the next conference in Missouri in 1994. Several concerns were expressed. Some thought the conference should be held later in the summer, perhaps mid to late June. Our seating was a little more cramped and not as convenient as at Montreal. We realize this, but initially expected a group of about 70 to 100 persons and had far more.

There was a desire expressed for some parent panels on personal experiences and ideas to use with MSUD children of various ages, including how older children dealt with the disease. (I think we all agree Elio Canella did a wonderful job expressing his perspective on being a teenager with MSUD.) Many parents of older children expressed surprise at advances in research and the new ideas that were presented.

Thank you for participating. We truly enjoyed meeting the families and sharing information. It was a joy to see all of you, and we hope to see you in 1994.

COMMENT FROM A PARENT

Details

Written by Sandy Bulcher

Published: 14 July 2009

Regarding the 1992 Symposium, our family had a wonderful time! The Sullivans and Toths obviously worked very hard in preparation for the Symposium and it showed. It is really special to watch the fellowship that takes place between the children. It is also great to be able to communicate about MSUD where people know what you are talking about (instead of the usual confused response that we are all used to). It struck me that talking about MSUD there is like talking about the common cold to the general public. I was able to get several specific dietary questions answered, which proved to be very helpful.

Dr. Menkes: It was fascinating to hear the process that he and his associate went through to determine the source of the maple syrup smell of the urine. Everyone felt a sense of gratitude toward him for all that he has done for MSUD.

Dr. McInnes: He mentioned that some doctors believe that the level of keto acids is what causes brain intoxication instead of the leucine level. Also, it was interesting to learn how much the leucine level can vary from patient to patient and even with the same illness and the same patient. (We are starting to find this is true with our son, Jordon).

Dr. Elsas: He suggested that if we are traveling, we can contact his office to find out where

other metabolic centers are located. I hadn't realized that children with MSUD could lead a fairly normal life with 5-10% enzyme functioning. The leucine oxidation test was interesting and hopefully will be helpful to all of us.

FIELD TRIAL AT SYMPOSIUM

Details

Written by Louis J. Elsas, M.D.

Published: 14 July 2009

Dr. Louis J. Elsas and Dr. Klein brought equipment for a study project for which they were looking for volunteers. It was a painless test but not totally tasteless according to the look on some of the children's faces as they drank their 'tasteless' drink. Dr. Elsas wrote the following summary of this project for our Newsletter. He will share with us the final results when the study is finished. This was a simple way for the families to aid in research. Thanks for the opportunity, Dr. Elsas.

The Toronto field trial testing children's ability to oxidize leucine-1-13C to 13CO2 was a great success. Basically this study was designed to answer the following questions:

- Can we measure whole body leucine oxidation with a single oral drink of leucine and analyze how much is expired as CO2?
- Can we differentiate degrees of impaired branched-chain αketo acid dehydrogenase among children with MSUD?
- Can we identify carriers of MSUD among siblings of patients with MSUD?
- Can we determine which patients with MSUD will respond to pharmacological doses of thiamine by sequential analysis of whole body leucine oxidation to expired CO2 'on' and 'off' thiamine?

Background

Dr. Klein and I had worked on a stable isotope method (13C) to study whole body leucine oxidation using intravenous infusions, multiple blood sampling, as well as sampling breath. Although this clearly identified the whole body's oxidation potential for leucine, it was too invasive (too many blood sticks) to make it clinically useful. Only 20% of branched-chain ?keto acid dehydrogenase is activated in cultured dermal fibroblasts and since most of the body's branched-chain amino acids are catabolized by liver, muscle, heart, and kidney, we need a better method to assess whole body leucine oxidation. Hopefully such a method could be used to answer questions 2, 3, and 4 and predict the degree of dietary restriction of branched-chain amino acids in babies with MSUD.

Results

Twenty-eight families and forty-six children volunteered for the non-invasive (no sticks)

breath test before the Toronto meeting. Affected children with MSUD had reduced amounts of 13CO2 recovered in their breath tests compared to their unaffected siblings. We have incomplete data as yet, but it appears that the more severe the requirement for dietary restriction, the lower the 13CO2 recovery in the child's breath. Therefore, this test has the makings of a method to predetermine how much dietary restriction of branched-chain amino acids will be required. Among eight children whom we tested 'on' and 'off' thiamine here at Emory, three clearly had increased whole body leucine oxidation 'on' thiamine while five did not. The three 'thiamine-responders' also had a clinical response and required less dietary leucine restriction 'on' thiamine.

We are still evaluating whether the carrier state can be determined in unaffected siblings by this breath test. The preliminary data says, 'yes.' It is clear that obligate heterozygous parents have reduced whole body leucine oxidation compared to adult controls. We do not yet have enough age matched normal childhood controls.

So, in summary, the preliminary answers to our four questions are, 'yes'. We need more clinical information on those of you who have not completed the clinical survey form and to develop more childhood-aged control data.

Thank you very much for your hospitality and enthusiasm. I look forward to working further with all of you to best care for our kids challenged by MSUD.

SUMMARY OF SYMPOSIUM 1992, TORONTO

Details

Written by Various Authors

Published: 14 July 2009

This was our 6th biennial family-sponsored Symposium. Forty-five families attended, many bringing their children with MSUD and their siblings for a total of fifty-five children. Other family members and professionals brought the total to 189. The Symposium was well organized. The atmosphere was comfortable and the food ample. The baby-sitting service kept the children supervised and entertained. Comments were positive.

The Agenda:

Friday a.m.- June 5

Welcome & Update on Ontario Association for MSUD Research *from the Toth and*

Sullivan families
The host families extended a warm welcome.Ê They explained the association they established to help fund the Symposium and MSUD research. Each person had been given a green cap with the Ontario Association logo during registration and were encouraged to wear them. Mike Sullivan faithfully wore his cap while moderating the conference in a lively manner.

Opening Comments and Thoughts on MSUD: The Disease *by Dr. Roderick McInnes, M.D., Ph.D., F.R.C.P. Associate Professor of Pediatrics and Medical Genetics, Hospital for Sick Children, Toronto, ON*
Dr. McInnes, in discussing genetics, told us everyone carries 8 to 10 genes that could cause severe diseases. He described the treatment and home management of MSUD. There are many variations of MSUD in patients, Dr. McInnes explained, and several unknown factors, such as which blood levels of BCAA are safe and what factors cause brain intoxication. He summed up three things he learned from his experience with MSUD÷listen to the parents, small details are critical (like weighing food), and advances will be in small steps (with the possible exception of gene therapy). Our goals should be to develop our group, support genetic research, stay sophisticated÷don't go for the quick fixes, and make your physician listen.

Two-Year Correlation Study of 2,4-DNPH, Keto-stix, and Blood Amino Acid Levels *by Doreen Anderson, Nurse Coordinator, Hospital for Sick Children, Toronto, and Alice T. Mazur, R.N., P.N.P., Division of Metabolism, Children's Hospital of Philadelphia, PA*
Alice Mazur stated that intellectual outcome for persons with MSUD depends on the age of diagnosis and levels of chronic control. A pilot program for newborn screening in PA began in several counties in the spring of 1991 and is to be extended statewide later. Chronic control involves a monthly analysis of leucine blood levels plus a dietary leucine analysis and early intervention in illness using DNPH urine testing. Doreen Anderson gave data on 39 DNPH urine testings. There were puzzling variations in the test results. The small sampling made it hard to draw any conclusions.

Teen Perspective on MSUD *by Elio Cannella*
Elio, a teenager with MSUD from Ontario, told his story. He was born March 12, 1974 and diagnosed at 13 days of age. He spent 2 weeks in intensive care and 3 months in the hospital. He has cerebral palsy and did not walk until he was 3 yrs. old. He planned to enter thirteenth grade this fall, studying computer graphics. He plays piano, likes wrestling, and has twin brothers who protect him. It does not bother him to have MSUD; he has accepted it and learned to cope with it. His friends accept him as he is. He always follows his diet, counting equivalents, but is angry at the formula. Watching other people eat doesn't bother him. Since he has not tasted many of the foods, he doesn't miss them. At eighteen he is doing quite well.

Reflection of MSUD with Case Histories *by Dr. John Menkes, M.D., Professor Emeritus of Neurology and Pediatrics, University of California School of Medicine*
It all started in Boston in 1952. Doctors had just learned about the effects of a restrictive diet in preventing retardation in PKU. Dr. Menkes had just finished medical school when he became interested in a mother who had the 4th baby with a peculiar smell in the urine. The other three had died and at 14 days of age, this one did too. Dr. Menkes described his long, arduous task of trying to learn the cause of the odor. It was a fascinating story of

determination and perseverance. The first child with MSUD was treated in 1957 by Selma Snyderman. Dr. Menkes concluded by mentioning there are twelve different forms of MSUD known to date, and there is a possibility of gene replacement within 5 to 10 years.

Low Protein Products *by Specialty Food Shops and Kingsmill Foods, Ltd.*
This was a description of two non-profit food shops in the Toronto area. Kingsmill special food items were introduced and displayed. Samples of a number of low protein foods were available.

MSUD: Current and Future Prediction and Therapy *by Louis J. Elsas II, M.D., Professor of Pediatrics, Director of the Division of Medical Genetics, Emory University School of Medicine, Atlanta, GA*
Dr. Elsas stressed the importance of early intervention in MSUD involving four components: screening, retrieval, diagnosis and treatment. Screening began in 1978 in Georgia. Eleven cases were found by Dec. 1991÷37% are African American. He explained his special research project which would involve some of the children at the Symposium. (See Field Trial at Symposium.)

Maternal Dietary Issues in MSUD *by Sandy Van Calcar, R.D., Metabolic Nutritionist, Weisman Center, University of Wisconsin*
Sandy gave an account of Sue Ann McKnight's medical care during her pregnancy. Although Sue Ann is considered to have classic MSUD, she ordinarily tolerates more protein than most other classics. She was the first woman with MSUD to give birth to a child. She delivered a normal, healthy baby after a very closely monitored pregnancy. Her protein and carnitine needs were much greater during pregnancy but returned to her normal level of tolerance after delivery.

Panel Discussion and/or Question Period
Parents kept four professionals busy answering questions. The panelists were: Dr. Elsas, Sandy Van Calcar R.D., Alice Mazur R.N., P.N.P., and Doreen Anderson R.N.

Saturday a.m.- June 6

MSUD Family Newsletter Update *by Joyce Brubacher & Peter Shaffer*
Peter explained some statistics about the organization. Joyce encouraged sales of our MSUD cookbooks and forgot all the other things she meant to talk about!

Low Protein Bread & Rolls Baking Demonstration *by Shayla Brubacher*
Shayla explained how to use an Hitachi bread making machine. With the help of a friend, she demonstrated the machine by putting in the ingredients and serving fresh warm bread 4 hrs. later. She had some previously made low protein pretzels and bread sticks to sample.

Teen Workshop held *by Doreen Anderson and Helen Phillips in an area adjoining the conference room*
This was an opportunity for the older children with MSUD to discuss and compare diets and experiences. They learned that all persons with MSUD share a love for potatoes, their

number one diet staple.

Psychological Testing: What Does It Mean? *by Jo-Ann Finegan, Ph.D., Clinical Psychologist, Department of Psychology, Hospital for Sick Children, Toronto*
Little has been written about psychological testing of persons with MSUD, possibly because individual medical centers have so few patients. Tests are yardsticks and psychological assessment is more than just test results. Dr. Finegan explained testing in general.

Dietary Workshop: Feeding the MSUD Child--Parents and Professionals Together *by Anna Marie Schaefer, R.D., MPH., Pediatric Outpatient Service, University Hospitals, Ann Arbor, MI*
Anna Marie covered these areas in this workshop: Model (diagram), Goals, Pediatric Neurology Clinic Experience, Parent Experiences, and Creating Variety in the MSUD Diet for School Age Children.

Language Disabilities and Speech Pathology *by Sue Scott, Speech Pathologist, Richmond Hill, ON*
Speech-Language Pathologists work with all age groups to improve clients' speech, language, and swallowing skills. Some Speech-Language Pathologists also work to improve feeding skills.ä So begins a paper passed out by Sue which covers much of her talk. This handout provides definitions of terms, gives risk factors for those with MSUD, explains speech, language and swallowing/feeding intervention, and what parents can do to encourage language development.

Humor in Chronic Illness Workshop *by Arlette Lefebre, M.D., Child Psychiatrist, Associate Professor for University of Toronto, Hospital for Sick Children, Toronto*
Dr. Lefebre gave a humorous talk with a serious message. Her handout covers how to build your own humor resources, organize and start using your resources, use humor for a variety of reasons, and develop humor resources at work. She also includes material on using computers with modems to access public forums.