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SYMPOSIUM 2000 PRESENTATIONS

Details

Written by Joyce Brubacher

Published: 20 July 2009

Latisha Love-Gregory, a genetics student at the University of Missouri-Columbia, attended Symposium 2000. She graciously offered to summarize selected presentations and submitted two summaries, one on MSUD treatment and one on research. Following these two summaries is an article on litigation involving newborn screening and an article on the highlights of the Adult/Teen Panel. The last article in this section "Parenting Leadership" was presented by Cristine Trahms, a registered dietitian.

PROGRESS AND UNDERSTANDING TREATMENT OF MSUD

Details

Written by Holmes Morton, MD

Published: 20 July 2009

Presented by Holmes Morton, MD
Dr. Morton's talk focused primarily on the effect of the disease on the central nervous system.

To illustrate the progress in the treatment of MSUD, Dr. Morton presented the case of a nine-day-old infant. The baby was referred to the Clinic because of a positive newborn screening test. The results were unfortunately late (as many parents have experienced first hand), and the baby was suffering from symptoms of Classic MSUD. However, within 36 hours of treatment, the baby's branched-chain amino acid levels had decreased significantly, and by 48 hours of treatment, the levels were within normal range. After 72 hours of treatment, the baby was taking formula by mouth, and her neurological exams had begun to normalize. By day five, she was ready to go home.

This was accomplished by metabolic management as opposed to the old method of dialysis. Metabolic management involves reducing the levels of the branched-chain a-keto acids and keeping patients hydrated so that these a-keto acids are removed from the body, thus reducing leucine levels.

Dr. Morton dazzled the audience with success stories that were the result of metabolic management. Of the 37 MSUD newborns that were diagnosed and treated by Dr. Morton at his Clinic in Strasburg, PA over the past eight years, 18 were diagnosed 12 to 24 hours postnatally. (This was possible because the parents had been tested and identified as carriers of the MSUD gene or had other children with MSUD.) With good metabolic management, none of these 18 babies became ill. The average hospital stay for Dr. Morton's patients is now 4 days compared to the months of hospitalization endured by patients and families years ago.

Understanding the systemic [pertaining to the whole body] management of MSUD improves treatment. This involves understanding the management of the many variables that make MSUD so complex. These variables include:

1. How to decrease leucine levels?
2. How to prevent valine and isoleucine deficiency?
3. What mixture of amino acids is needed to support normal protein synthesis at a rate that is optimal for normal growth and development?
4. What caloric intake is allowable?
5. What is the role of other amino acids in supporting a homeostatic environment?

One of the many challenges faced by Dr. Morton, along with other physicians and researchers, is understanding MSUD's effect on the brain. Knowing the level of leucine is important, but it is just as significant to determine the pattern of amino acid variation in general which leads to the neurological symptoms. To emphasize this principle, the audience was given a quick lesson in neuroanatomy. Dr. Morton analyzed and interpreted MRIs and amino acid profiles from the records of several patients with metabolic disorders who were battling cerebral edema.

While treating these patients battling edema, Dr. Morton and his colleague, Kevin Strauss, made a significant observation: the severe edema was very sensitive to extracellular serum osmolarity and their sodium level was related to the brain edema. Dr. Morton concluded that the increased leucine concentrations block the export of other amino acids which require a sodium-dependent transporter for moving efficiently across the cell membrane. This causes the cells to take up more water and swell.

Understanding the effect of MSUD on the brain involves more than understanding cerebral edema. Doctors and scientists need to look deeper and explore the relationship of leucine, valine, and isoleucine to other amino acids essential for the normal functioning of the nervous system. Therefore, the continued progress in the treatment and management of MSUD relies heavily upon diagnosing these patients as newborns in order to develop a better understanding of the interrelationship of leucine, valine and isoleucine to the remaining essential amino acids.

GENETIC AND BIOCHEMICAL APPROACHES TO MSUD

Details

Written by David Chuang, PhD

Published: 20 July 2009

Presented by David Chuang, PhD

Dr. Chuang's talk focused on MSUD research performed by his laboratory and colleagues. Although MSUD was first reported about 46 years ago, Dr. Chang stated, "There is still no cure for MSUD, but progress has been made in biochemistry, molecular genetics, and in the treatment of the disease." Dr. Chuang attributes this achievement to the tremendous advances made in research over the last 15 years. His talk focused on several aspects of MSUD research.

One aspect is the **biochemistry of MSUD** which involves studying the interaction of the biomolecules which constitute the Branched-Chain a-keto Acid Dehydrogenase (BCKD) complex and their relationship to other cellular components. The BCKD complex plays a vital role in the catabolism of leucine, isoleucine and valine. It was quite exciting to hear that Dr. Chuang, along with his colleagues, had resolved the crystal structure of the branched-chain a-keto acid dehydrogenase. This is the first report of resolving the crystal structure of any human a-keto acid decarboxylase. The highlight is that, with this knowledge, researchers could now predict more confidently how *certain* mutations may interrupt the normal functioning of the BCKD complex and cause MSUD.

The **molecular genetics** (studying and identifying the genes which encode the BCKD complex) and the **mutational analysis of MSUD** are tasks that can be tedious when identifying new mutations that lead to MSUD. There are six different genes that encode for the BCKD complex; however, only 4 of them have been fully sequenced at the DNA level. To further complicate matters, the four genes are located on different chromosomes in the genome, and a mutation in any one of them has the potential to cause MSUD. As Dr.

Chuang explained, mutational analysis is very important. To emphasize this, he listed several points:

1. Identifies mutations for prenatal diagnosis of MSUD
2. Sheds light on the association of a mutant allele with certain groups
3. Facilitates the study of the biochemistry of MSUD
4. Promotes the development of biochemical strategies which can be used to mitigate the disease

Dr. Chuang also commented that in order to use gene therapy, doctors first need to know which of the patient's genes is affected. But, as he informed the audience, *"This is not as simple as it sounds."*

Dr. Chuang discussed thiamine responsive MSUD. After several studies, his lab has observed a strong correlation between the thiamine-responsive phenotype and a subset of Type II MSUD (mutations in BCKD E2 component). This suggests that patients in this category are candidates for thiamine treatment.

The study of **assembly defects** involves utilizing biochemical strategies to determine how the proteins that constitute the BCKD are shaped and folded, as well as, how the proteins potentially interact. From his extensive studies with cell cultures, Dr. Chuang was able to conclude that the defects in protein folding and assembly, caused by certain MSUD mutations, can be partially reversed by specific chemical compounds. Dr. Chuang stressed that this is just the beginning, and these observations depend greatly upon the nature of the defect which causes MSUD.

NEWBORN SCREENING AND LITIGATION

Details

Written by Joyce Brubacher

Published: 20 July 2009

By Chuck Hehmeyer

Chuck Hehmeyer is an attorney from Philadelphia who is very concerned about the number of children who die or are damaged because they were not picked up by newborn screening. He presented "Newborn Screening's Potential Litigation" at Symposium 2000. Rather than printing a summary of that talk, here is an article he wrote for this issue of the Newsletter.

Newborn screening (NBS) in the United States has been a great success on one hand, yet a great public health failure on the other. It's wonderful to see most kids with PKU and congenital hypothyroidism (CH) growing up healthy today. But it's sad that children with so

many other disorders that benefit from early intervention (like MSUD, MCAD, GA1, Galactosemia, etc.) often are not screened.

Quiz question: How many states in the U.S. currently screen for MSUD? (The answer is at the end of this article.)	I represent a beautiful little girl in Texas with classic MSUD who sat in a coma in the ICU of a major hospital from day seven of life until day 21 while lab studies were "pending." It is a miracle she is even alive. Of course she is now profoundly delayed. Her doctors were confused by her presentation (mistaking it for sepsis). NBS should and could have prevented the heartache of this little girl, her family *and* her doctors. But Texas doesn't screen for MSUD. Why?

Most of us know that Robert Guthrie pioneered NBS in the U.S. Frustrated by early opposition to PKU screening from the American Academy of Pediatrics (AAP), Dr. Guthrie and PKU parents went straight to the state legislatures. (It's true, the AAP opposed PKU screening in the 1960s.) Their compelling presentations resulted in legislated PKU screening in all 50 states, saving scores of children over the next 35 years from a lifetime of disability. Unfortunately, this prompted legislatures to delegate NBS oversight to state departments of health, resulting in *state employees* deciding the future progress and development of NBS. Since then, doctors have almost completely abdicated their responsibility for thinking about metabolic screening to public health officials.

This situation is unique to NBS and really a historical quirk due to Dr. Guthrie's decision to go directly to the state legislatures. Doctors, rather than government, continue primary oversight of other medical screens like pap smears, mammography, prostate exams, glaucoma testing, hearing exams for newborns and children, etc.

While we now have uniform screening in the U.S. for PKU, CH and sickle cell disorders, NBS varies dramatically from state to state for the 30 or so other metabolic disorders that (1) benefit from early intervention and (2) are difficult or impossible to diagnose before a child is harmed. **Let's call it what it is: newborn roulette.**

Today we know that tandem mass spectrometry reliably and inexpensively identifies more than 20 disorders (including MSUD) using the same blood spots Dr. Guthrie pioneered - which are obtained from *every* U.S. newborn. But few states use this technology. State departments of health have proven to be poor managers of NBS systems for familiar reasons. State health officials resist innovation and protect state screening jobs - at any cost - from competition from private labs that do the screening more accurately and less expensively. Even when states themselves invest in tandem mass spectrometers, they engage in what amounts to on-the-job training for the first	Left foreground: Melissa and Jessica Berman with John Devantier (All three with MSUD). Right foreground: Samson Li and daughters (Edlecta in seat has MSUD).

few years (suffering unnecessary false negatives) rather than just awarding state screening contracts to experienced private labs like NeoGen in Pittsburgh.	Back left: Kathryn Burkholder (cured of MSUD with liver transplant) and center, Austin Sprock (MSUD) held by grandmother. Photo taken at Symposium 2000.

So what can we do? Obviously, parents and support groups make great advocates. I encourage all parents to lobby state legislatures, departments of health, and even the media, to demand that screening be expanded to all disorders that benefit from early intervention. Additionally, parents of children harmed by delayed diagnosis of a controllable metabolic disorder should know that it may be possible to redress this wrong in the legal system.

The first question parents usually ask is whether we can sue the state for not screening or force the state to begin comprehensive screening. The answer is almost universally no. State entities are immune from suit, particularly for what the courts view as discretionary legislative and/or executive action. We live in a democracy, and lobbying and voting are the appropriate remedies for perceived mistakes in policy making by government officials. [State laboratories can be sued in some states for mistakes in the screening and reporting process itself (e.g., Ohio: yes, California: no), but the courts will never permit lawsuits to answer what they view to be executive/legislative policy questions like *what* metabolic disorders to screen for.][1]

Although we cannot sue states themselves for failing to offer comprehensive NBS, in some states we can sue HMOs and hospitals for failing to screen. I have filed three such suits - two in Pennsylvania and one in Texas. There is an interesting split in the U.S. Some states permit suits alleging that the whole medical profession lags behind in adopting new technology. Some don't. States like Pennsylvania, Washington and Texas permit suits against medical professionals for not adopting clearly appropriate technology - even if *none* of the other healthcare providers in the state offer the technology. The argument is: hospitals and HMOs cannot insulate themselves from liability for failing to keep up with important developments simply by uniformly failing to use them. Other states (e.g., Ohio) allow medical professionals to set their own standard of care exclusively, no matter how unreasonable. These states say, essentially, if all the hospitals do it, it's okay legally - no matter what. (By the way, no other group gets this special treatment - except doctors.)

We've made the most headway on newborn screening by suing doctors (pediatricians, neonatologists, ER and NICU doctors, etc.) for delay in diagnosis of a sick child with a metabolic disorder. When doctors realize that they wouldn't be in this position if the child had been screened, *they* put the heat on. In Pennsylvania, we now have over 95% *voluntary* participation in supplemental newborn screening using tandem mass spectrometry.

Answer to the quiz question: As of July 2000, only 20 states in the U.S. screen for MSUD - less than half.[2] If the little girl in Texas with MSUD had been born in Georgia or Alaska, she would go to normal school today. But she wasn't, and she's not. Ivory tower public health officials have no problem justifying this. But they could never explain how this makes sense to the family of that little girl. I doubt they could explain it to twelve ordinary people on a jury.

1. This is another strong argument in favor of opting for screening by private laboratories rather than state labs. State labs that are immune from suit do not have the strong economic incentive that private labs have to get it right.
2. *Pediatrics*, August 2000, Volume 106, No. 2 (supp.) p. 393.

TEEN/ADULT MSUD PANEL SAYS, "YOU MUST DRINK AND DRIVE!"

Details

Written by Joyce Brubacher

Published: 20 July 2009

Trish Mullaley presided over the Teen/Adult Panel as they fielded questions from the audience. She shares an account of the panel discussion.

The teen/adult panel in the background. From the left: Mark Silva, Katy Foster, Lauren Codner, Leanna Peters (hidden), Vanessa Funes, Emily Talley, Jeffery Fredricks, Melissa Berman, and Shayla Brubacher. In foreground to the left is Nikolai Rudd and to the right is Michael and Sharlene Woorman. All of these are teens and adults who have MSUD.	You must drink your formula if driving. This message was heard crystal clear at this past MSUD Symposium 2000. Teenage and adult individuals with MSUD stressed very strongly the importance of drinking formula before getting behind the wheel of a car. The drivers said that lack of formula makes them feel tired and less alert and indicated that no matter what, formula must come first. This includes during sporting activities, at school and anywhere requiring full attention. Having MSUD does not seem to have a negative impact on sports. Those individuals with MSUD, who are also involved in sports, seem to have enough energy providing they drink plenty of their formula.

It seems that all of these bright young ladies and gentlemen are well aware of their disorder and the consequences that follow if they do not comply. When asked if any of them had ever cheated on their diet, the response seemed uniform. Cheating is really not an option since their leucine level will elevate making them feel almost drunken or lethargic. While all admitted to having done *some* cheating, they also indicated that they didn't feel well and felt guilty; therefore, cheating just doesn't seem worth it. Their perception of "cheating" is consuming larger amounts of leucine than their daily allotment.

When asked if their friends know about their having MSUD, most responded that they do. Responses were mixed when asked just how much their friends know about the disorder. Some are not shy and are of the opinion that they really don't care what others think. Their friends seem to understand the seriousness of the disorder, and, therefore, watch what their MSUD friend eats at a party and watch for odd behavior. Others, who are more introverted, tell their friends only what they need to know. They keep things simple. Their friends know that they have dietary restrictions for medical reasons, and that's it.

All of them seemed to understand things from a parent's perspective and showed respect and concerns for parents raising a child with MSUD. While many admitted that they didn't like having their parents hovering over them when they were younger, in hindsight, they seemed to grasp just how scary it must be for a parent to raise a child with a disorder that can be life threatening.

This was a wonderful, articulate group of young men and women. Kudos to them and their parents for a job well done!

- Trish Mullaley

MORE PARENT COMMENTS ON SYMPOSIUM 2000

Details

Written by Joyce Brubacher

Published: 20 July 2009

- "The speakers were great, so much information and insight. I really loved Dr. Morton's talks. He gives all of us great hope."
- My first meeting with Dr. Morton was very satisfying. I like the way he explained the information. I have learned in two days more than I have known in 25 years. I have so much to learn, it is like I need to start at point one because things have changed

so much with the new technology and new understanding. Every family that has a PKU or MSUD child should attend one of these Symposiums. They are very educational and you get to meet a lot of wonderful people."

- "Things I learned: (1) Leucine levels going from 48 to 5 mg/dl in 36 hours - that is advanced management of MSUD! (2) I should be using the tyrosine the doctor gave me for my children with MSUD that is sitting in my cupboard! (3) Valine deficiency makes children (and rats) irritable. (4) Most of the cortex formation of the brain takes place before 1 yr. of age and this depends on correct nutrition to the brain."
- I really enjoyed Dr. Morton's discussions. He is able to bring the complexity of MSUD down almost to my level. I liked the update about newborn screening, Sandy's discussion, Neo Gen, the attorney and Harvey Levy. I also liked talking with all the families. I would like to get more information about Neo Gen. I think I am going to start giving out information about how to get this testing done to people I know who are pregnant. - Denise Pinsky
- The last conference I was at was four years ago. It's wonderful to see how nicely these young men and women and children have developed. - An Aunt
- I learned two new things: Being constipated and spending too much time in the Jacuzzi can potentially elevate leucine levels! - Sandy
- As I sit here looking over our group, I have to think, MSUD is definitely not a respecter of persons - it can be anyone or anywhere in the world. It unites us and we feel for each other and understand each other. We can also be thankful for all those older ones that "paved the road" for these younger ones. But their brain damage will not disappear; it will be with them the rest of their lives. Much is being learned and that is why we have these Symposiums. It is helpful to us just to see all the families together and to hear each speaker. - Edna Newswanger
- Because I have an older child, I didn't have much interest in newborn screening, but after listening to the speakers on this subject, I have gotten more interested. I would even like to help with newborn screening on a volunteer basis. Also I got a lot out of the short but informative speech about the Scott C. Foster Metabolic Research Fund. I enjoyed and found very informative the breakout for the preteen/teen/ parent group. The "Gene Repair Therapy" talk by Dr. Morton was very good. - Renee Eck

PARENTING LEADERSHIP: THE MOVE FROM COMPLIANCE TOWARD SHARED MANAGEMENT

Details

Written by Joyce Brubacher

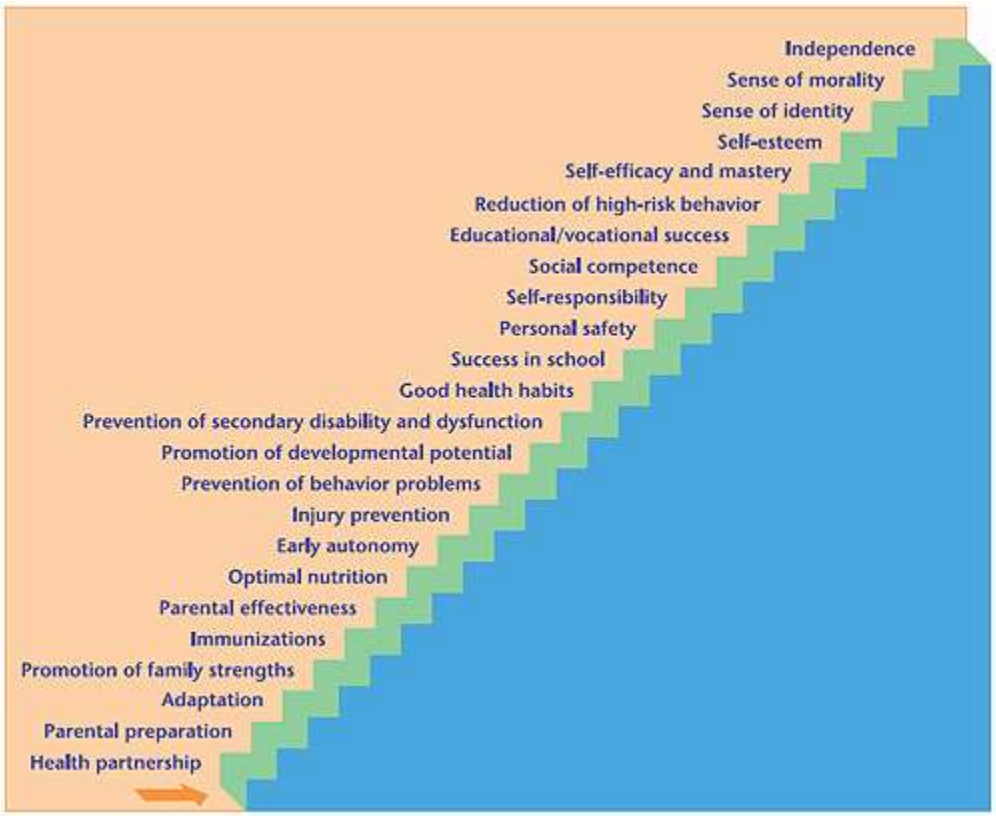
Published: 20 July 2009

Presented by Cristine M. Trahms, MS, RD, FADA
Cristine Trahms, from the Department of Pediatrics, University of Washington Seattle, presented these models and guidelines for managing a chronic condition - such as MSUD. This material also appeared in Pediatric Nursing 2000.

Children with chronic conditions encounter all the typical challenges of growing up in addition to special ones that stem from their condition and its management. However, overall health outcomes remain similar despite the presence of the chronic condition. These outcomes are well detailed in the Bright Futures model (See graphic). The model begins with the development of a therapeutic alliance when the child is an infant and progresses through a continuum of social, developmental, and health outcome achievements that lead to an independent, healthy adult.

Children with chronic conditions should achieve these same health outcomes.
Children and youth ideally progress through appropriate developmental stages to become knowledgeable and capable participants in their own care. Typically this developmental process parallels development in other areas of the children's lives. When successful, the process enables children to share in the management of their health condition and to achieve the outcomes articulated in *Bright Futures*.

Bright Futures Health Supervision Outcomes
Central to the concept of health supervision is the belief that specific preventive and health-promoting interventions lead to desired outcomes. The social, developmental, and health outcomes summarized below contribute to the overall health and well being of infants, children, adolescents, and families. These outcomes occur along a continuum, varying in their timing from child to child and family to family.



Taken from *Bright Futures: Guidelines for Health Supervision of Infants, Children, and Adolescents,* M. Green, Ed. Arlington, VA: National Center for Education in Maternal and Child Health, 1994. http://www.brightfutures.org

Developmental models underline the concept that learning is based on cognitive readiness. The developmental approach also provides a *forward* momentum for the child's self-management skills and responsibilities. Parents monitor the child's progress and emotional status and support the child's efforts and negotiate changes in responsibility as appropriate. By *working with* the child, rather than *doing for* the child, the parent comes to know when the child has mastered skills at the current level and is ready to move on.

The second model, the Leadership model, shows the direction of shifting responsibility for care of the chronic disorder from parent to child. It depicts by bold arrows the need for the parent to guide this movement and the important idea that both parents and child have a role to play. (See graphic)

The Leadership model indicates the dynamic nature of the parent-child relationship during effective management of a chronic disorder. Initially, the parent provides all of the necessary care of the child, regardless of the child's age. As the child matures in cognitive and physical skills and gains experience in managing the condition, the parent transfers some of the responsibility for self-care to the child. The parent becomes the *manager* and the child the *provider* for these carefully articulated skill-appropriate responsibilities. The parent is available to support the child's *provider* skills and stands ready to re-assume some of these tasks

for a short time if it is necessary because the child is ill or other life complications require additional parent support.
(LEADERSHIP MODEL GOES HERE) put heading The Leadership Model

As the child becomes more confident and competent in self-management, the parent and child negotiate the next step. The parent becomes the *supervisor* and the child becomes the *manager* of specific tasks. The parent is, again, poised to resume the managerial role or specific tasks for a short time, if necessary. However, the parent and child must both understand and agree that the child should not be allowed to regress to a previous stage. The parent provides additional support during times of stress, and the child continues to develop self-management skills and responsibility.

Eventually, the parent assumes a *consultant* role in the child's management of his/her disorder and the child becomes the *supervisor*, manager, and provider of care. The parent supplies information, support, decision-making guidance, and resources, but the child assumes ultimate responsibility for his/her health care as the *CEO*.

The question remains: How do families keep focused on the *big picture* of their child's general growth, implement the actions necessary over time to move their child forward in shared management, while remaining ever mindful of the condition specific needs of their child? Successful families operate by changing the focus to immediate medical needs or to normative family life depending on the *current needs* of the child and family.

Each child with a chronic medical disorder has a component of specific immediate medical needs that must be interwoven into normative individual, family, and community life. During times of wellness, these specific medical needs may appear to be in the background and remain relatively out of focus. They are, however, present and always able to be brought back into clear focus by the family if the need should arise.

The models require bringing together many components into an effective dynamic system that is day-to-day family functioning. Some of the components can be *systematized* so that they function as integral parts of *normative* family or community life and thus effectively guide management of the chronic condition so that it remains in *normative* rather than *crisis* focus. Some systematic approaches that have been reported as effective are:

1. **A system of monitoring of the condition.** Parents who are successful develop a *system* for monitoring the disorder that is integral to family daily life and activities. Some successful system components are (1) the weekly structured family meeting; (2) disease management tasks integrated as a part of daily family tasks, i.e., the family *chore schedule* indicates take out the garbage, brush your teeth, drink your "milk," pack your lunch for school; (3) a notebook is kept on the kitchen counter for recording food eaten or other activities related to medication (notes are added as events occur).
2. **A system for involving the child at skill level.** The tasks that are the child's responsibilities are based on the child's physical and cognitive capabilities, e.g., a toddler may count the number of crackers for lunch while a third grade student

   may prepare formula independently on Saturday morning.

3. **A system for evaluating the child's success/errors and remediating these errors.** Parents and family members support success by pointing out to the child evidence of successful management as it occurs, such as consumption of formula without a reminder, as well as the natural consequences of poor management, such as developing strategies to insure that after school formula is consumed.
4. **A system for celebrating small successes.** If something is a two-stage process, celebrate each step along the way rather than just the final outcome. For example, celebrate the child's remembering to select the correct snack foods at a surprise party for a friend, as well as maintenance of a low weekly branched-chain amino acid level.
5. **A system to prevent parental burnout.** Parents need ongoing support for themselves since management of the disorder is a lifelong parental and child activity. Support of the child, emotional or physical, doesn't stop at age 10, or 13 or 18 years. Successful children have parents who remain involved in a qualitatively different way but do remain involved in the child's life. Parents who are able to remain consistent and supportive of their child but also negotiate the direct management role based on the child's skills and needs do not suffer from "parenting burnout" as readily as parents who assume the total and on-going responsibility for the child's health and health management.

Effective parental support of a child with a chronic condition requires a thoughtful parenting strategy. Parents do not have the luxury of casual or inattentive parenting. They are required to be focused and anticipatory in their parenting style if their child is to successfully accomplish both the developmental tasks of childhood and disorder management.

Actions that Support Leadership Skills

Stage/Age	Child Capabilities/Actions That Form the Basis for Leadership Skills	Parent's Leadership/Actions to Support the Child's Growing Capabilities
Infant (0-12 months)	Though dependent on parents for care, it is helpful if the child gives clear cues of distress so parents can grow in the recognition of emergent needs and make appropriate responses For example, clear cues of hunger and satiety help the parent understand when to offer formula and when to withdraw it	Learn ramifications of the condition and how/what resources can help Learn how to ask questions that can assist managing the condition in the context of an overall healthy living pattern Participate in support activities to increase knowledge of disorder and its management

	Clear cues of optimal health (adequate growth, development, and social interaction) also enable parents to identify the positive impact of their actions to promote good management	Develop routine regarding daily treatment that fits with family life patterns Recognize signs of immediate distress and seek emergency care Recognize signs of early distress and seek evaluation Learn to acknowledge those challenges that are developmentally typical for most children versus challenges specific to the child's condition Learn how to share information with extended family and daycare providers See/acknowledge evidence that the child is thriving under attentive management Assume the role of "repository for condition specific information" regarding the child's reaction to the treatment
Toddler (1-3 years)	Cooperate with routine treatments Help hold equipment and work with parent to make equipment function as needed, for example, use of blender to prepare formula Develop a sense that parents are a source of help/comfort	Develop rituals regarding treatment so child knows what to expect and can begin to learn through repetition Begin to recognize that the child needs to have roles in the management of the condition

	Accept constraints of condition and treatment with limited behavioral acting out, for example, "yes foods/no foods" Understand firm limits of parents, for example, "no"	Identify possible roles the parents are willing to begin to share with the child Change the established management routine based on the child's growing capabilities and areas of cooperation Continue to build clinical and community support network
Preschool (4-5 years)	Identify body parts important to early identification of a problem or treatment Test limits of cooperation Magical thinking may lead to fears Imitate adult's behaviors Learn labels for condition specific "problems" in order to communicate treatment needs Learn labels for feelings associated with condition and its treatment so can communicate feelings	Acknowledge regressions, allow very brief period of reorganization and then resume and praise prior skill performance Set fair and appropriate limits Model acceptance of the management routines and limits Encourage some flexibility in rituals of treatment so child begins to experience multiple ways to accomplish same goal Develop relationships with school personnel regarding specific needs
Early School Age (6-9 years)	Recognize and act on 1 or 2 of major internal body cues of a problem Actively participate in concrete monitoring of condition Increase understanding of condition, i.e., cause and effect, a concrete level of what's going on inside the body to necessitate management	Continue to label cues and give positive reward for child's recognition Start negotiating with child for what each party will do regarding management and set criteria for forward movement that fits with family life

		Be prepared to re-negotiate for cause! Establish logical consequences for actions Negotiate the "rules" for working together to get all necessary treatments completed Be positive and reinforcing about what needs to get done Support normative activities and integrate treatment needs Model telling others about the disorder for the child Discuss the approach to telling teachers, friends, coaches, etc. about the disorder and the amount of detail necessary to share
Late school age (10-12 years)	Increased level of understanding of condition - begin to understand long-term needs Use labels that are medically correct in order to effectively discuss with providers Learn how and when to respond to peer pressure yet still take care of self Enact most psychomotor skills associated with treatment with parental support Learn more sophisticated system for reporting symptoms, management steps, outcomes	Remain present for the child that is involved in care and monitoring decision-making Accept the manager versus CEO role in much of treatment Insure child has told important others: friends' parents, coaches, etc. of the condition and what assistance they could provide if needed "Be there" in case of emergencies and new aspects of disorder

	Develop specific set of self-management tasks that are completed independently	Provide the tools so the child can self-manage (get the formula, get the prescriptions) Support the child in actively communicating with their provider Encourage discussion of the child's monitoring system so as to help them grow in understanding
Early adolescent	Main manager of daily, routine care Develop strategies to complete all of the necessary routine management tasks Know how to effectively ask for assistance in complex situations Know where can be flexible vs. not flexible and be able to enact the flexibility when appropriate	Shadowing of parent activities Negotiating and re-negotiating of who does what. Becoming the consultant versus remaining the manager Discuss new issues (sex/drug/alcohol) for their normative and any special condition effects
Late adolescent	Make a commitment to lifetime treatment Increase understanding of the disorder and its long term as well as short term consequences on other aspects of life-vocations, intellectual achievement, well being etc. Sense of self as capable manager of disorder Integrate the realities of the condition with the invincible nature of their years	Develop a flexible way of communicating with the youth in order to stay informed while not seen as interfering Remain present for support and problem solving with the youth Provide support and guidance as the youth transitions from pediatric to adult care services

	Appreciate benefits that the constraints of management allow Continue to develop more independent clinic and community support network as transition to adult-based care services	

C.M. Trahms and G. Keickhefer, "Chronic Illness in Children: Supporting the Development of Children as They Move From Compliance Toward Shared Management." *Pediatric Nursing*, 2000.

MSUD Management Timeline

Age of Child	**Tasks for Children and Parents**
0-6 months	Parents learn about and adjust to MSUD
6 months	Start low-protein solid foods
6-7 months	Introduce cup
8-9 months	Introduce finger foods
10-15 months	Consider weaning from bottle (discuss transition with clinic staff)
2-3 years	Learn the concept of "formula first" Learn to distinguish "yes" and "no" foods
4-5 years	Begin to learn to count foods - "how many" Begin to use scale - "how much"
5-6 years	Assist in formula preparation Teach children how to deal with other children's curiosity about MSUD

7-10 years	Prepare formula with decreasing supervision Choose after school snack Learn to pack school lunch Begin to list foods on food record Begin weighing food regularly on scale
10-12 years	Begin to prepare and consume formula independently each day (with parental monitoring) Prepare simple entrees independently Know what blood levels are ideal
13-14 years	Increasing self-monitoring (with continued parent support) in formula preparation and consumption Independently manage total leucine intakefor the day Learn menu planning Responsible for food records
15-17 years	Responsible for all aspects of self-management Able to do "finger poke" for blood test Able to explain basics of MSUD - "What is it?" Responsible for remembering recent blood levels Continued parent support
18 years	Transition to adult-based clinic care Ready to live independently, including: - formula preparation and consumption - food preparation and records - blood tests for serum BCAA levels as requested

University of Washington, 2000

FOOD NEWS - CAMBROOKE FOODS

Details

Written by Joyce Brubacher

Published: 20 July 2009

Shayla Brubacher, holding an energy bar,	Lynn Paolella, owner of Cambrooke Foods, supplied free samples of her energy bars and bagels at Symposium 2000. They were a big hit with parents and children. Both items contain less than 1 gm of

and Sharlene Woorman with Lynn Paolella, of Cambrooke Foods.	protein. The 4 oz. energy bars have 39 mg of leucine and the 5 oz. bagels contain 24 mg of leucine. The bagels and energy bars can be ordered on-line or call 508-276-1800. They will soon have a toll free number and plan to add other products. Keep updated on this exciting new company by checking their web site: www.cambrookefoods.com

FOOD NEWS - NEWS FROM SHS NORTH AMERICA

Details

Written by Joyce Brubacher

Published: 20 July 2009

New product: SHS North America is pleased to announce a new product, **Loprofin Drink Mix**, a milk substitute! One 14 oz. can of the powder makes more than 5 quarts of Drink Mix. It can be added to **Loprofin Cereal Loops** or used in everyday cooking and baking. This new product will be available in early 2001. Call 1-888-LOPROGO for free recipes with your order.

New packaging: Beginning in 2001, **Loprofin** will have a new face! Throughout the first months of the new year, you will see the Loprofin brand in new, attractive packaging with easy-to-read nutrition information. No changes have been made to the products themselves.

Discontinued: SHS North America is going to discontinue **Loprofin Canned Bread** in the spring of 2001 (items #TWL 501 and TWL 503). Please place your orders by early spring. For more information, visit their web site at www.shsna.com or call 1-888-LOPROGO (567-7646).

NEWS & NOTES - NATIONAL COALITION FOR PKU & ALLIED DISORDERS

Details

Written by Joyce Brubacher

Published: 20 July 2009

This organization, not to be confused with the New England Connection for PKU and Allied Disorders Inc. in Massachusetts, is becoming very active and influential. Included with this issue is a registration form for their first big Metabolic Conference to be held in Dublin, Ohio. The agenda is on the back page of this issue.

The National Coalition for PKU and Allied Disorders Conference is planned for families and professionals from seven different groups: MSUD, PKU, the organic acidemias, glutaric aciduria, tryosinemia, homocystinuria, and the fatty oxidation disorders. During the day on Friday, each group will have its own speakers and agenda. On Friday evening, there will be a Newborn Screening meeting open to anyone interested in that topic. There is no cost to those who attend only Friday evening.

On Saturday, the topics will be of general interest to all attendees. On Thursday and Saturday evenings there will be receptions at the hotel with cheese and crackers, fruit, and a cash bar for those who want to attend. Be sure to make reservations soon at the Dublin Embassy Suites Hotel. The toll free number is 1-800-362-2779 or call the hotel directly at 614-790-9000.

Be sure to state that you are attending the National Coalition for PKU and Allied Disorders Conference in order to get your $99 room rate. You will have a choice of 2 double beds or 1 king size bed. Each room has a pull-out couch, microwave, refrigerator, coffee maker, blow dryer, etc. This price includes a continental breakfast and an early evening snack and soft drink. There is a $50 registration fee which includes lunch both days.

There is an indoor pool, whirlpool, and small exercise room. They ask that children with MSUD under 12 not attend the conference as there is no program or daycare planned for them. For more information on this event or organization, check out their web site: pku-allieddisorders.org

NEWS & NOTES - ADDITION TO CLINIC

Details

Written by Joyce Brubacher

Published: 20 July 2009

A full time pediatrician, Dr. Kevin Strauss, will join Dr. Morton and the staff at the Clinic For Special Children in Strasburg, Pennsylvania this summer. The Clinic is expanding to make room for the increase in staff. On December 29, with the help of many volunteers, a timber frame was raised for the addition.

The following paragraph from the Winter 2000 issue of the *Clinic For Special Children Newsletter* describes the work of the Clinic.

Unlike most centers which specialize in genetics, the Clinic was designed not only to provide specialized diagnosis, laboratory work and consultation, but also to provide primary medical care to children for all their medical needs however minor or critical. This comprehensive approach has given the Clinic an opportunity and advantage of learning more about disease processes related to metabolic disorders through observation in many different circumstances and timely intervention for more effective treatment. When we moved into the present building, we thought we would see and treat children with a dozen to two dozen different genetic conditions. To date we have seen and treated children with over 80 different genetic conditions. Our work has led us toward a greater role in teaching others about what we have learned. We hope with the expansion in staff and facilities we will be able to do more in sharing progress through lectures, publications and conferences and in educating students of medicine.

The expansion of the Clinic is a key part of a five year plan which includes a second (and potentially a third) pediatrician who specializes in rare genetic disorders and the growth of an endowment fund to give the Clinic financial security. The Clinic has a non-profit status and relies on donations as well as proceeds from three annual auctions. This keeps fees for patient services at a minimum rate since most of the families who use the Clinic do not have insurance and do not participate in federal or state aid programs.

Another paragraph from their Winter 2000 Newsletter states the reason for the changes and the Clinic's goals for the future.

During the next year we are having a carefully planned and needed growth spurt. We are building . . . building staff, building space, and building the Clinic's future through the endowment fund. We do not plan to grow like this every year or to grow so that it changes the fundamental and personal way in which we operate and serve children. But, we need to grow in a way that we can continue to provide state of the art medical care, laboratory

services, and [to] support research and education that is based on the needs and experiences of our patients.

NEWS & NOTES - ARTICLE ON GENE THERAPY

Details

Written by Joyce Brubacher

Published: 20 July 2009

Those who are interested in the gene repair therapy that Dr. Blaese spoke to us about at Symposium '98 will find an article worth reading in the May 1999 issue of *Fortune* Magazine. "Can Gene Therapy Cure This Child?" reviews the Kimeragen company's struggle to break into the field of gene therapy. The article chronicles the problems involved in bringing the therapy to human trials. The Floyd Martin children with Crigler-Najjar disease, who are scheduled to be the first to receive the new gene therapy, are featured in the article. It is a lengthy and interesting article although MSUD is not mentioned.

NEWS & NOTES - FOLLOW-UP ON FENUGREEK TEA AND THE SMELL OF MAPLE SYRUP

Details

Written by Joyce Brubacher

Published: 20 July 2009

In the Spring/Summer 2000 issue of the Newsletter, a short article stated that fenugreek tea may cause an odor in the urine similar to maple syrup. We receive many calls from persons who smell maple syrup in their own urine or on their child and wonder if they could have some form of MSUD even though they are not sick. Recently a woman, 39, who had raised a family, was diagnosed with intermittent MSUD. However, she had been very sick several times prior to the diagnosis. Are there many more undiagnosed cases?

After reading the article on the fenugreek tea odor, Dr. Neil Buist responded with a letter to me providing some additional information. I certainly appreciate his response, which follows.

The most common "MSUD" question that I usually get is about "healthy" children who smell of maple syrup (MS) as mentioned in the Spring/Summer issue. Almost all of these children are developmentally normal. Clearly, fenugreek is not the cause in most of these children, and indeed, it is not common to smell MS in children or adults who eat lots of fenugreek in curries.

I think that the smell (which derives from some unknown compounds) comes from what older clinicians used to call the "second liver" - the bowel. Gut bacteria are responsible for making certain essential nutrients and there are many types of bugs, either aerobic or anaerobic, that live in the gut. Many are not even identified! Not everyone has the same collection of bacteria nor is the population constant all the time. It has been shown that the adult bowel contains about a half pound (220 g) of living bacteria; what a metabolic powerhouse!

Anyway, I think that the MS smell comes from some of the anaerobic bacteria in the gut, which explains, of course, why there are no changes in the plasma amino acids or the urine organic acids in these cases.

- ***Neil M. Buist, M.D.***

NEWS & NOTES - JOIN THE EXCLUSIVE MSUD EGROUP

Details

Written by Joyce Brubacher

Published: 20 July 2009

Emily Talley is a 43 year old adult with MSUD. She has set up a group e-mail account for anyone who would like to use it as a forum for group discussion concerning MSUD support.

The group e-mail account is absolutely FREE. It's hosted by egroups.com, and only members can access it. The fact that it is a member only group e-mail account makes it a safe place for people to receive and give support around the issues of MSUD.

For those of you who aren't familiar with group e-mail accounts, here's how they work. When you post a message to the group's e-mail address (msud@egroups.com), your message will automatically be sent to every member in the group. Likewise, when someone responds to a member's e-mail, their reply will automatically be sent to every member in the group.

Group e-mail accounts are a great way for a group of people who share common concerns/interests to have on-line discussions. They allow for very rapid exchange of information between a large number of people.

Emily Talley is the owner/moderator of the MSUD group e-mail account. She can be reached at emily@inherhands.com. She will trouble-shoot and fix any technical difficulties that come up (e.g., bounced e-mails, etc.).

To become a member of the MSUD group e-mail account, just send an e-mail to: msud-subscribe@egroups.com

NEWS & NOTES - NEWBORN SCREENING CAMPAIGN & UNITY QUILT

Details

Written by Joyce Brubacher

Published: 20 July 2009

Tyler for Life Foundation, Inc., a newborn screening awareness organization, recently launched a national campaign, **"Handout America,"** during which volunteers supply pamphlets entitled "A Parent's Guide to Newborn Screening." These pamphlets are to be displayed in hospitals, doctors' offices, clinics and any place that serves children and families.

The organization claims that each year there are 2700 children in the U.S. who die or are brain damaged but could have been saved with a newborn screening test. If you would like to volunteer to distribute the pamphlets, use the order form on the Tyler for Life web site or e-mail Kileen Hall at kileen@tylerforlife.com giving your address and the number of pamphlets you want. Ten pamphlets per doctor's office is the suggested maximum.

The Tyler For Life Foundation will be collecting quilt panels for a **NBS Unity Quilt** from families with children who have disorders detectable through newborn screening. This new project will involve all the families in a unified effort. The quilt will allow the families of children with these disorders to celebrate and share the lives of their children.

The mission of the quilt is to create a visual testimony of the most compelling symbol of newborn screening - the children touched by it. The quilt will be unveiled on Sept. 1, 2001 in celebration of NBS Awareness Month. To participate or learn more about the project, visit the Tyler for Life web site. You can also learn more about newborn screening, related disorders, and the Tyler for Life Foundation at tylerforlife.com or call 1-888-454-3383.

The Tyler for Life Foundation also hosts a discussion listserv for persons interested in corresponding with others involved in newborn screening. To join, send an e-mail to: NBSAdvocacy@listbot.com

NEWS & NOTES - TAX DEDUCTIONS FOR CONFERENCES

Details

Written by Joyce Brubacher

Published: 20 July 2009

The IRS will allow deductions for some travel expenses when parents attend medical conferences for their children. On May 8, 2000, responding to a request from Congressman George Miller, the IRS issued a ruling that will allow parents to deduct some of the costs associated with attending medical meetings related to their children's health condition. Parents will be able to deduct "amounts paid by an individual for expenses of admission and transportation to a medical conference relating to the chronic disease of the individual's dependent." See IRS Bulletin 2000-19 for more information: http://ftp.fedworld.gov/pub/irs-irbs/irb00-19.pdf

NEWS & NOTES - MCDONALD'S SALADS

Details

Written by Joyce Brubacher

Published: 20 July 2009

Greg and Ann Fredericks checked into the possibility of getting garden salads at McDonald's restaurants without the egg and cheese included on the new shaker salads. According to the McDonald's web site, they are willing to make adjustments to "make you smile!"

The Fredericks received a letter saying it should be possible to get a made-to-order garden salad at any McDonald's. They suggested calling 30 minutes ahead of time so they have time to make it. However, their local McDonald's said they don't need to call, and they will only need 5 to 10 minutes to prepare it.

The Fredericks have requested the salads when traveling and usually have had no problem. Sometimes the employees refuse and need to be reminded that it is possible to make a special order. It is best to ask only when the McDonald's is not busy as it does require time. Better than the older type of garden salad, the shaker salads have greener lettuce, small tomatoes, and scallions.

So check with your favorite McDonald's and make arrangements to have your request honored. Have your salad, keep your diet, and SMILE!

FAMILY HISTORIES - OUR GIFT FROM HEAVEN

Details

Written by Joyce Brubacher

Published: 20 July 2009

Cheryl Trudeau

At first it was like a fairy tale dream. Little did we know it would soon turn into a bad dream and a never-ending nightmare from which I just couldn't wake up.

My story begins on September 23, 1985, the day I gave birth to my first and only son, Shayne Warren Davis. His weight at birth was 6 lbs. 6 oz. and he was 19 inches long. He had big brown eyes with some gorgeous, blonde hair. He seemed to be a perfectly healthy baby boy with all ten of his fingers and toes.

Shayne was born at Valley Medical Center here in Fresno, California. My delivery was normal with no complications, so everything seemed to be fine. We were released from the hospital the next day and moved in with my parents until we could find our own place. I tried to breast feed him from the moment he was born, but he refused to nurse. So after two days of trying, I gave up and went to bottle feeding. I tried Similac formula, but he would only drink an ounce at a time every three hours. I was really starting to become concerned when his appetite kept decreasing at each feeding.

On the fifth day, he was down to drinking almost nothing. By the end of the day, he had only consumed three ounces. His last feeding was a half ounce at 11:35 p.m. that night. On the morning of September 29th, I awoke at 7:35 a.m. Shayne had slept through the night without waking for any feedings. I went to his crib, and he was lying in a very odd position with his back arched. He was stiff and seemed to be in a very deep sleep. I tried to feed him, but he wouldn't open his mouth. I couldn't open his mouth with my fingers; it was like his mouth was glued shut. I got really scared at that point.

I took Shayne to the emergency room at V.M.C where he was born. A doctor came in to examine him right away. He shook his head and said he would be right back. Two more doctors examined him, and they too were very confused about what was causing these symptoms. Then Shayne started having seizures, one after the other. They gave him massive doses of phenobarbital and finally brought the seizures under control. They admitted him to this hospital where he was born. The doctors thought he could be diabetic, but a blood test ruled that out.

He was taken to the Newborn Intensive Care Unit and isolated from the other babies. Shayne was now in a coma and was put on a respirator to help him breathe. His condition was getting worse; his kidneys became weak, and they feared he would have kidney failure. The doctors knew that the longer he stayed on the respirator, the weaker his lungs and other vital organs would become.

After two weeks, the doctors decided to try to get him off the respirator. They tried twice, but he had to be revived each time. They told us it was very hard on his little body each time he was revived, and asked if I wanted them to try again if he failed to breathe. Did I want to put him through that kind of trauma again? I was so upset and unsure of what to say, all I could do was cry.

I was given a little time to decide. Once I calmed down, I began praying, asking God to please give me the wisdom and strength to make this major decision. And He did. I knew I had to think about what was best for Shayne and not for my own selfish reasons. It was the hardest decision I have ever had to make, but it was also the best. I told the doctors that if my son didn't breathe on his own, they should let the Lord take him and let him be at peace, feeling no more pain. I was only seventeen and had to make a decision that would change my life.

The doctors went to his room and began the process of taking Shayne off the respirator while I stood outside his room and prayed. I begged God to please allow my baby boy to live on here with me for as long as possible. I kept saying, "Breathe baby Shayne, please breathe." He stalled for a few seconds, and then sucked in a few breaths of air and began breathing on his own. The doctors were so surprised. They came running out of his room saying, "He did it, he really did it!"

I was totally shocked, but happier than words can say. My son did it; he had such a strong will to live. God really did hear my prayers, and from then on I had faith in God. Before then, I wasn't sure there really was a God.

Now it was time to really focus on finding the cause of his critical condition. After several more doctors examined Shayne and were unable to make a diagnosis, a specialist in genetic metabolic diseases was called in. That's when I met a very special doctor and person, Dr. Susan Winters. In my eyes and Shayne's, she was heaven-sent.

Shayne was about three weeks old by then. Dr. Winters examined him and the first thing that she noticed was the odd smell from his urine. It was a sweet smell, like maple syrup. Dr. Winters had never actually treated, or even seen, a patient with MSUD. She had only read about it, and Shayne had all the symptoms. She drew blood and sent it to the nearest lab which was in San Diego.

It seemed like it took forever to get the results back. When they finally did, the results were positive. My son had maple syrup urine disease. We wanted to know all about this rare disease with such an odd name. We had no idea what we were going to experience. Dr. Winters consulted with another doctor from somewhere back East for advice. The first thing she had to do was to figure out the right mixture for Shayne's MSUD formula. It took several tries before she got it right. I don't remember exactly what it consisted of, but a little bit of regular milk was added, and, for a brief time, thiamine was added. They had to tube feed him for a short time until he would suck from a bottle.

Shayne was finally able to come home at almost five months of age. It was the beginning of February, and he did pretty good. He got sick a few times after that, but it wasn't too bad. He was also hospitalized a few times for different reasons. Since then, Shayne had surgery on his legs to correct the damage done while he was in the coma. The surgery was not very successful. Even though the doctors aren't very hopeful, we still keep on hoping that some day Shayne will walk.

Because California does not test all newborns for MSUD, my son suffered severe brain damage. Shayne is now 14 years old but has the mind of a 4 to 5 year old child. He has three sisters who are all healthy--Heather 12, Cassie 5 and Frances 3. He seems to love

them all as much as we love him. He has brought us so much joy. Shayne lives with my parents, Warren and Brenda Goodman. They take great care of him, and I am still a big part of Shayne's life.

Thank you for allowing me to share my story.

- Cheryl Trudeau

UPDATE: *This history was received last year. Shayne has had a total of three surgeries on his legs. None have proved very successful. Currently he is getting therapy for his legs and also struggling with a reflux problem. If California would have had a comprehensive newborn screening program at the time Shayne was born, today at 15, he might be enjoying normal teenage activities. California still does not screen for MSUD*.

A Special Thanks to a Very Special Doctor

I hope my son's doctor is reading this because it really comes from my heart. I would like to say thank you to a very special person, Dr. Susan Winters, from here in Fresno. Dr. Winters, I just want you to know how much my family and I thank God He sent you to us in 1985 when my son, Shayne, was given no hope of living. You were there for me during the most difficult time in my life. Not only are you an excellent doctor but a warm, caring and very loving person. Any patient would be very lucky to have you as their physician. So thank you Dr. Winters, you will always have a special place in my heart.

Your friend and more,

Cheryl Trudeau

FAMILY HISTORIES - MARIA FERNANDA AND HER FAMILY

Details

Written by Joyce Brubacher

Published: 20 July 2009

The following story was translated from a May-June 1997 issue of a metabolic bulletin published in Chile. The Aravena-Orellana family first contacted our organization in November 1997. They had heard about the MSUD Symposium planned for 1998 in Lancaster, Pennsylvania and were eager to attend. In a fax they told me they were collecting bottles to try to raise money to attend. With help from the MSUD Support Group,

Manuel and Sarita and their daughter Maria Fernanda were able to attend Symposium '98. Sarita and Maria attended again in 2000.

"The love of Sarita and Manuel was very quick." Within a year after they met, they were married and Maria Jos? was born soon after. Eight years later, Jos? Miguel was born. Then followed the next pregnancy which was received with great joy.

On January 22, 1991, after a lengthy caesarean operation, a beautiful blue-eyed baby girl was born. They called her Mar?a Fernanda. Seven days later, she started having feeding problems. She would throw up after eating. Treatment was started. She was not very active. A lot of people said she was "just a little lazy."

Months later, she overcame the feeding disorder. Her weight and height were normal for a girl her age. The concern was forgotten by everybody except Sarita, who because of her child-caring job, sensed that something was wrong--that her daughter was "different." Mar?a Fernanda appeared to be autistic. She loved to play with little wheels that she would make turn and turn. She regressed from all her little achievements and, little by little, she started to isolate herself from the world around her. She underwent many exams, but nothing precisely defined her condition.

Two years later, the family was connected with the organization, Parents of Autistic Children. They were advised that Mar?a Fernanda was not autistic. It was recommended that she attend a nursery school to improve her language skills. With this help, Maria started to talk and develop social skills. Although Maria Fernanda seemed to be happy, Sarita and Manuel still had concerns.

One day in December, pale as a sheet of paper, Maria was taken urgently to the Van Buren Hospital in Valpara?so. Dr. Daniel Abumohor suspected a metabolic disorder and contacted Dr. Marta Colombo, a specialist in metabolic disorders. Fourteen days later, she got a precise diagnosis: maple syrup urine disease.

Sarita, sitting on a sofa when she heard the diagnosis, felt as if she was starting to sink and anguish took over her heart. She could only cry and cry. She walked for long hours and found herself at home waiting for her husband Manuel. The news she had to tell him was so overwhelming. Now what? And tomorrow? And in two more years?

Manuel just cried. The dinner table was the center of family communication, and now, with all the dietary restrictions Mar?a Fernanda had to keep, it would no longer function as such. Mar?a Fernanda was a "special" girl, and the process of becoming a "special" family was beginning: to learn from the pain, to build constantly, and to close ranks in order to confront with fortitude and much love this new way of life. Unexpectedly, a new path was opened and hope reborn. The family contacted the MSUD Family Support Group and were nurtured by their experience and solidarity, their will power and affection.	

SHARING

Details

Written by Joyce Brubacher

Published: 20 July 2009

The MSUD Symposium 2000 was, as always, interesting, informative and moving. I would like to thank everyone who worked to make it happen (especially Herb Foster and Trish Mullaley) as well as everyone who participated.

After talking with many parents and hearing lots of horror stories, I wanted to suggest a way that we, as a group, might better help each other. It is apparent, unfortunately, that there are few doctors in the United States who have experience dealing with MSUD emergency situations.

Most of us travel as families, and, as our children get older, they will travel more often alone. In our case, we put a lot of effort into travel preparation, but things can still go wrong.

Just to give you one quick example: our son, Nikolai, went to visit his brother Thaddeus, who was living in Los Angeles at that time. Since a formula order was about to be processed two weeks before the trip, we asked our pharmacy to ship it directly to the Los Angeles address. Thaddeus confirmed receipt of the formula box, and Nikolai flew to L.A. carrying just one extra day of formula. This prevented his having to deal with heavy formula in baggage or lugging it onto the plane.

Nikolai arrived at his brother's apartment, opened the box and discovered that the wrong product was inside. By the time he called us, it was late in the afternoon here on the east coast and, of course, this had to happen on a Friday.

With just minutes before closing time for the weekend, we made frantic calls to the formula manufacturer, the distributor, the L.A. hospitals, and our doctor. We finally located a nearby L.A. pharmacy where formula could be shipped (they could not ship it to a private address), and our efforts proved successful - but not without incredible strain and pressure on us. It was a near disaster. Nikolai could have been without formula for three days.

It occurred to me during the recent Symposium that if that happened or some other MSUD crisis to a MSUD child or adult anywhere near where we live in western Massachusetts, we would know how to get help for them. We would know where blood could be drawn and spun, how to get overnight or even same day delivery to the nearest equipped hospital, and who could be trusted on a local level to consult with their primary doctor. We also know where the nearest doctor familiar with MSUD is located, and could perhaps lend some emergency formula from our kitchen if it happened to be the same type. In case of emergency, Barbara and I could also assist with advice since we have had more than 26

years of experience with various MSUD problems. I would think we could be of comfort to another family if their child was caught in a crisis situation.

We suggest every family connected with MSUD, who is willing to help other families, make a note of this type of emergency information to keep on hand for easy reference. On the next family address list, or in a separate mailing, all willing families could be identified. The information necessary to make a quick contact with a local family would be included for emergency assistance. Then when an MSUD adult or family travels, and there is a crisis, they would be able to check the family list and locate a nearby contact person.

Although it is important to encourage more doctors to specialize in this field, that will take time. In the meantime, if we can be of support to each other, it could ease some of the stress resulting from traveling away from our primary doctor and hospital.
Please contact Joyce Brubacher (editor) with an updated address, telephone number and e-mail address and indicate that you are willing to be a travel emergency contact family. But make sure you then jot down MSUD emergency information that could be used in your area and keep this handy.

- ***Eric Rudd***

Your clinic may also be able to supply emergency contacts in the area where you plan to travel