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FEATURED FAMILY - MSUD, A PARENT'S EXPERIENCE

Details

Written by Sandy Kiel

Published: 17 July 2009

Sandy Kiel presented the following account at the Association for Neuro-Metabolic Disorders (ANMD) Conference on April 16, 1994. Sandy wrote about their first child with MSUD for the Family History section of the Nov. '91 Newsletter. Although this report is not exactly a family history, it provides an interesting update on their family, so I am using it in this section. Sandy was speaking to a group of mostly PKU parents. She describes MSUD so well I reprinted the whole speech.

We are Carl and Sandy Kiel. We are parents of four children: Adam-9, Sara-7, Jenna-3, and Jesse-1. Our two youngest children have MSUD, so dealing with the disease and diet have become part of our lives. For those of you not familiar with MSUD - it is a metabolic disease in which the child cannot tolerate the branched-chain amino acids: leucine, isoleucine, and valine. MSUD differs from PKU in that it is so critical to detect in the newborn period to prevent brain damage and death. A child will get very sick very fast if not detected. MSUD gets its name because the urine smells like pancake syrup when levels are high. For someone who did not pay much attention in Chemistry and Biology, I have learned a lot!

Jenna is now three. She was born in 1991, the first baby to be detected by Newborn Screening in Michigan. Because our other children did not have the disease, we were surprised when informed that Newborn Screening showed our baby had MSUD. We truly believe it to be a miracle, because Adam and Sara were born before Newborn Screening began in 1987 and would not have had the advantage of early detection. In fact, Dr. Allen had told us that the state considered discontinuing MSUD screening, because they had not found any babies with MSUD previous to Jenna being born!

Dr. Allen confirmed Jenna's diagnosis and we started her on formula at 5 days of age. However, the newborn period is so critical and she was already showing serious signs; a high-pitched cry, fussy and irritable, loss of sucking reflex, and then, by 10 days, she was lethargic and nearly dead. We spent a week at the University of Michigan on TPN and she

did great!

Jesse is now just one. I had an amniocentesis to determine if he also had MSUD. Carl and I, together with Dr. Allen decided to have the test to provide the best care for Jesse right from the start. He never had high levels his first year until his recent illness in March '94. We brought the formula along to the hospital. Doctors, staff, and nurses were all alerted and very careful. Many nurses would ask about MSUD and were very curious because they were familiar with the newborn screening and never heard the results of it.

Jesse has broken all the records - he weighed 10 pounds at birth and has now nearly tripled his weight! He is not just thriving - he is the picture of good health!

DIET: Jenna and Jesse are both on formula (MSUD Diet Powder) that they must drink each day. Both are very limited in their protein intake, about 4-5 grams per day currently. This has fluctuated with their health. Jenna has to be coaxed to eat and drink, Jesse eats and drinks everything! I write down every day what they eat and at night compensate with milk to reach the desired level for protein.

HOME MONITORING: Like many other families, once a month we do a home monitoring blood test on the blood card and send in 3-day diet sheets to have Dr. Allen check their levels. I also have them weighed at our family doctor and record this monthly.

URINE TEST: We also have a urine test, DNPH. Equal parts of urine and chemical are combined. If their keto acids are elevated, their urine gets cloudy. The degree of cloudiness correlates to the level of keto acids. I do this when I am suspicious of their actions, or if they are ill. With MSUD the urine has the sweet smell of syrup. In Jenna and Jesse, they are usually showing other signs and quite sick before I notice the syrup smell.

When Anna Marie, [the Keil's nutritionist], asked me to talk today about illnesses I just laughed, because I would panic and call her when the kids would get sick! But we are getting a little better at it - I don't panic quite as quickly, but I do still call her!

When an MSUD child has a common illness, it is often accompanied by high levels and serious signs of MSUD. Not counting Jenna's newborn hospitalization, we have had four major bouts with illness - three for Jenna and one hospitalization for Jesse. It happens about once a year. We are very careful of flu and ear infections. At the first sign of any symptoms I go to the local doctor to have it checked and treated. Jenna had recurring ear infections the last two years. The doctor keeps a close eye on her and has her on a maintenance antibiotic. The flu has been the most serious illness resulting in hospitalization.

When our children are sick (flu or infection), I watch carefully for these signs - they are in progressive order:

1. Tired eyes - Jenna gets dark circles under her eyes and just looks exhausted at the first sign of any illness.
2. Change in their behavior-reduced activity or grouchy and crying.
3. DNPH test - I do this at the first sign of illness. The degree of cloudiness will increase as levels rise. The smell of syrup usually comes later for our children.

4. Ataxia (loss of balance) - this can be slight, like not being able to sit up with the support of a hand or tripping a lot. Or it can be as serious as suddenly unable to sit or stand at all!
5. Lethargic - very tired with little or no activity.
6. Blood levels - these are taken during an illness and will show a dramatic increase in leucine levels. Often the child is well and looking fine and leucine levels are still elevated but drop dramatically.

These steps can progress very fast - this could all happen in the matter of a day or less!

Our family doctor checks for common illness and stays in contact with Dr. Allen. We hospitalize based on the child's behavior or need to go on an IV due to dehydration. This decision is made by our family doctor together with Dr. Allen.

When we hospitalize locally, there are a few things we have learned. The first is what Jenna needs and doesn't need. We try to eliminate unnecessary tests and procedures. They are listening to us!

The second thing is to have the doctors keep in contact with Dr. Allen. Our local hospital has worked well with him for the most part. We often are educating the resident doctor as to what MSUD involves, and we tell him what to do and what is needed. They ask many questions. We feel like we tell them what to do - we just don't have those Dr. initials!

The last thing is we learned is to speak up when we don't agree with what the doctors want to do. We know our child and the disease better than they do. We have learned you really are your child's best and sometimes only advocate!

During Jesse's recent hospital stay, I explained the home monitoring blood test to the doctor. He said, "So then you're are a nurse?" I said, "No, I'm just a parent who's been well educated."

I keep a journal for both Jenna and Jesse. I record each month's weight, blood test results, and food intake. When they are ill, I keep a detailed account of what they were like each day, what we did, and the blood levels and DNPH results. Looking back at the journals really helps me and I see similarities in their illnesses.

In some ways, dealing with MSUD has made a monumental change in our lives, and in some ways it's now just a way of life we take for granted. When Jenna or Jesse get ill, we are reminded of just how fragile their lives are. Adam and Sara have become experts in MSUD. They've explained it to all their friends and educated the neighborhood kids. At meal and snack times, we always try to give Jenna something similar to what we are eating, if she can't eat what we have. We try to make it no big deal and fit it into our lives.

I want to share one cute story. Vern Ehlers was the state senator from the Grand Rapids area. He introduced the legislation for MSUD newborn screening and helped to get it passed. When Jenna was born, we sent him a letter and thanked him for his work. This fall, when he ran for US congressman, his office called us and asked to use Jenna's picture and story in their campaign brochure. We agreed, and Jenna was so proud of her picture she would show everyone and point it out! We thought it was a really neat opportunity to share

our story.

In closing, like many other families we feel we owe so much to Dr. Allen, Anna Marie, and all the staff. They have really educated us so that we know what we are dealing with, and that we are comfortable with it. They are always there and always supportive. They treat families with great respect and they are so dedicated to our children. Thank You!

SHARING - JORDAN'S CHICKEN POX EXPERIENCE

Details

Written by Sandy Bulcher

Published: 17 July 2009

I was feeling fortunate that Jordan hadn't been exposed to chicken pox. About that time big brother Tyler broke out with them. He was very sick, including a fever and poor appetite for four days.

I couldn't imagine how Jordan's levels were going to stay somewhat normal if his experience was anything like Tyler's. I called my pediatrician, who promptly called in a prescription for Zovirax. She said that the medication would lessen the fever and other symptoms. She was hopeful that Jordan would be able to get by with a few trips to the ER for IV fluids. That sounded good to me, because I was feeling as though I might as well pack my bags for a few days at Children's Hospital. I also called a few trusty MSUD moms for some insight, which proved helpful.

As the end of the incubation period grew nearer, I checked Jordan frequently for the dreaded spots. He understood that the chicken pox might affect his levels, but he wasn't concerned.

Monday, March 6, I noticed the first few spots. I immediately went to the pharmacy to pick up the Zovirax prescription, as it is most effective if started in the first 24 hours. The next day, Jordan had a low grade temperature, but was eating and drinking well. Dr. Allen had asked that I check Jordan's urine DNPH twice a day. I also did a finger stick and sent it to Dr. Allen.

Days two, three, four, and five went without incident. Jordan's appetite wasn't affected at all. He could drink all of his formula and I decreased his daily leucine intake only slightly. He had no further fevers and minimal spots from the pox. His urine DNPH remained negative and leucine levels remained 1.0 to 1.6 mg/% (his normal range).

Unfortunately, I had wasted energy worrying about Jordan's bout with chicken pox for no reason. Anna Marie, Jordan's dietitian thought that it might be helpful for others to learn about our positive experience. I feel that the medicine, Zoviraz, careful monitoring, and a few prayers from friends allowed for the best experience possible.

SHARING - ATTENTION DEFICIT DISORDER OR MSUD?

Details

Written by Anna Toth (Mike's mom)

Published: 17 July 2009

I have often wondered if I was the only MSUD parent who many times had to actually take my child's face in my hands and talk up close to him to get his attention or to stop him from talking continuously. Was it a normal thing for a child this age; or was it high levels; or Attention Deficit Disorder (ADD)? I always thought my husband suffered from it because he could never hear me when I asked him to do something, or if I was talking about my day! He just suffers from "Selective Hearing Disorder," and I will write a paper on it some day.

I turned to other MSUD parents to see what they had experienced. I found that in the pre-kindergarten and kindergarten age, there seemed to be very little concern about the disorder. I don't think I thought much about my own child during those years. Kindergarten is a very social based time with emphasis on motor skills, group cooperation, and basic cognitive learning. I did not worry much, as he could build with blocks, sing a song, color and paste, etc. Kindergarten is a fairly fast paced and less structured setting than in the primary grades. It isn't until primary grades, and with more structure and organization and less activity, that difficulties begin. Children are expected to sit and concentrate on reading, math, numbers and the alphabet, or story writing for longer periods. This is when many parents responding to my inquiry found their child could quickly fall behind if they were unable to concentrate or sit still.

We all seem to know that when levels are running even slightly higher than normal, we can see some hyperactivity and less concentration in our children with MSUD. In this respect I know that it can cause problems in school, because until the diet or illness is corrected, the problem is there. Many of us find that after an illness it takes a week or more to get the children settled back into a proper learning routine. At times they may have to relearn portions of their work because of the setback. This seems a form of ADD linked directly to MSUD and its control. Hence the importance of good diet control and constant blood monitoring.

I also heard from parents who have tried Ritalin for diagnosed ADD and are getting marvelous results in school. They report positive effects on their children's school work, such as having them stay on a task and carrying through their responsibilities without too much reminding or "nagging." A concern about Ritalin is that it suppresses appetite, and with MSUD that can be a real issue. It is important to work on correct dosages with your professionals so there is no weight loss. As one professional put it; you should try the recommended treatment for ADD. If it does not help, you will know a chemical imbalance is not the cause of the hyperactivity. You may have to look elsewhere for the reasons.

Some parents feel the MSUD is responsible for ADD; others feel we tend to blame too much on the disorder. It is very easy to allow more to "slide" with children with MSUD, because they already have so many restrictions in their lives. The stressed or tired parent may find it taxing to constantly remind or repeat instructions when the child seems not to hear them. These children require so much extra attention, it can be frustrating to follow through so often with simple instructions. It is easier to let it go, which can lead to behavioral problems.

So, what is the answer? I am inclined to think that only you can answer for your child. If there is concern, have the child tested. If you find your child meets many of the criteria for ADD, you may wish to try Ritalin. Work closely with your doctors and teachers. Keep your child's levels in the normal range and maintain good control of the diet. Each child with MSUD is unique, and you know your child best.

Thanks to the parents who took the time to responded to my request for your experiences with ADD. In particular, to Mrs. Carl (Marty) Zimmerman, who would be willing to share their experiences with ADD, hyperactivity and the use of Ritalin.

I hope you all have a wonderful new year in 1995. I will probably spend part of it pondering this MYSTERY THAT IS MSUD! Whatever did I think of before my son came along? I'd ask my husband, but he never listens anyway!

SHARING - SYMPOSIUM EXPERIENCE

Details

Written by Renee Eck

Published: 17 July 2009

What I expected to get out of the MSUD Symposium:

- a better understanding of TPN.
- more information about DNPH.
- more information about home managing.

From talking to other parents, I felt I wasn't doing all I could to keep my son Cory (who is 9

years old) out of the hospital. Cory has been admitted to the hospital over 30 times. Six to eight of those admissions were for surgery. He has had three hamstring releases; other surgeries were for putting tubes in his ears. After every hamstring surgery he was released from the hospital, only to return within 1 to 2 days, with a new set of problems.

Cory's doctor didn't seem concerned about these events, and never gave me any more information about other things I could do at home to help Cory stay out of crisis. When asked by another parent about using DNPH and TPN, he would not give a positive response and sometimes you got no response at all.

Even with all the information I received at the Symposium, I needed a physician to help me use these treatments. This problem was also solved at the Symposium. Dr. Helen Berry from Cincinnati, Ohio was one of the speakers. I live in Lancaster, Ohio, about a 2¸ hour drive north of Cincinnati. I talked to Dr. Berry a number of times at the symposium. Sandy Bulcher, another parent from Ohio, and I had the same doctor for our sons. We told Dr. Berry about this doctor whom we shared for 3 years. I explained the things that had happened to Cory these past 9 years.

Dr. Berry helped find me another doctor in the state of Ohio. She even went the extra mile by calling the doctor and letting her know all about Cory and the problems we have had. She was so very helpful.

This being my first MSUD Symposium, I felt I picked the best one to attend. I got all my questions answered, plus a new doctor to help me with using DNPH, Home Management, Sick Formula (which I never heard of before) and TPN if the need would ever arise to use it.

We are now seeing Dr. Nancy Lesilie, with whom I couldn't be more pleased. I had some questions about some medication Cory was on, and she sent him to a neurologist to get those questions answered. Cory has since had some medication changed and is now getting over some side effects that the other mediation caused. He is doing much better. He didn't go into the hospital the last two times he became ill. Because of better home management and "Sick Day Formula" (which we call "Super Milk"), Cory has become a lot easier to handle day to day. His personality has changed and his problems at school have improved due to the changes in his medication.

I feel I got so much more out of the Symposium than I could ever dream of. I finally feel I am doing everything I can to help Cory get as far as he can in life. It was well worth the time and money.

I want to thank everyone in Missouri for planning and organizing a great Symposium. I also thought you would like knowing the positive effect it had on our family's life.

I hope this next Symposium in Ohio can be of as much help to at least one family or child as this one was to me. Thanks so much.

SHARING - MSUD WORKSHOP

Details

Written by Sandy Bulcher

Published: 17 July 2009

This workshop was part of the Association for Neuro-Metabolic Disorders (ANMD) Parent Conference held in Ann Arbor, MI.

Dave and I and our two boys attended our first ANMD meeting Saturday, April 16, 1994. We met first with four other MSUD families and dietitian, Julie Jacobson. As always, exchanging ideas, sharing feelings, and getting to know each other better was so special.

We discussed several different topics including school issues, illness, and dietary information. We agreed it is so important that teachers are aware of our children's special dietary needs and signs and symptoms of increased levels. Unfortunately, some teachers are less receptive than others, and it's important to seek out one that will work well with you and your child.

I shared my occasional feelings of powerlessness over MSUD, especially after my son Jordan's recent illness and hospital stay. We discussed how stressful it can be when our children are ill, yet how important it is to maintain control over our feelings for the child's sake. Like other children, MSUD kids seem to catch a variety of illnesses from ages 2-5. Joyce Brubacher emphasized that frequent testing of urine DNPH, along with antibiotics at the onset of unusual symptoms, helped to minimize episodes of increased leucine levels for her children. She also shared that it's easier to reason with older MSUD children about the importance of formula, fluids, and calories during time of illness. Sandy Kiel recently learned that the chicken pox vaccine should be available this Summer.

We were excited to learn that Ross Labs has a new MSUD Food List, which Julie will send to each of us. (One M & M candy has only 5 mg of leucine. Yeah!) We discussed "sneaking" foods and agreed that for some younger children it may be beneficial to keep "no" foods out of reach. Also, if children test their parents by sneaking "no" foods, a matter-of-fact response might be appropriate. We really enjoyed the fellowship that we shared with the other MSUD families and look forward to our next gathering.

DR. ROBERT GUTHRIE HONORED

Details

Written by Joyce Brubacher

Published: 17 July 2009

Wayne and I were surprised and pleased to be invited to a dinner given in honor of Robert Guthrie, Ph.D., M.D. It was held in Buffalo, NY on April 24 by the Heritage-Oak Foundation Third Founders' Award Celebration. We accepted the invitation to show our appreciation for the way his work has benefitted our family personally and other families with children with MSUD.

The event honored Dr. Guthrie's work in preventing mental retardation for some 30,000 individuals worldwide through his initiation of the process of neonatal screening for inborn errors of metabolism. This honorary dinner was also a fund raiser for the Heritage-Oak Foundation. Dr. Guthrie has been a member of Heritage Center for more that 35 years and past president of its board of directors.

Dr. Guthrie began his career as a cancer research doctor, but became interested in preventing mental retardation after his 6 year old son was diagnosed with developmental disabilities. He devised a simple, inexpensive test for Phenylketonuria (PKU) in the early '60s and stimulated thinking in terms of preventing brain damage. It opened the field of neonatal screening which now includes other genetic disorders like MSUD. All states in the US now require the PKU test for newborns and many other countries have adopted screening programs.

Although PKU tests proved very effective in early trials, Dr. Guthrie worked diligently to overcome the reluctance of physicians who opposed laws requiring the tests. The tests were set up in public health departments and some doctors saw them as a threat to private practice. Parent groups in the US and in other countries were influential in publicizing the value of the tests. Dr. Guthrie believes the combined efforts of parent groups and professionals can be very effective. He and his wife have been advocates for people with mental retardation and are active in various parent organizations associated with mental retardation.

In 1963 Dr. Guthrie developed the bacterial inhibition assay for leucine that has since been used to screen for MSUD. This could be administered along with the PKU test. In July '64 five PKU screening laboratories located in western New York, Massachusetts, Oregon, New Zealand and Los Angeles began field trials using the test for MSUD.

Oregon had the only statewide program at the time our son Monte was born in Portland on March 12, 1965. Being the first in the world to be detected through a state neonatal screening program, he made history. Dr. Guthrie was in California at the time, but came to Oregon to see the test. We still have the article from the Portland newspaper, The Oregonian, which includes a picture of Dr. Guthrie looking at Monte's test.

Little did we understand the significance of this test at the time.

Very little was known about MSUD in the '60s. Without the Guthrie test, Monte's diagnosis would certainly have been delayed, if he was diagnosed at all. When Shayla was born in Indiana in 1970, the sate had no screening test. Would she have been detected early if Monte had not been diagnosed? Dr. Guthrie's dedicated work has been a tremendous blessing to our family.

Sometime in the mid '80s I was reading about Dr. Guthrie and Wayne and I discussed calling him. I would not have hesitated had I known how personable he was. When I said my name, he immediately asked if I was Monte's mother÷approximately 20 years after his birth! He also offered to call back so I would not have the phone bill. This interesting chat began our friendship.

Dr. Guthrie expressed great interest in our organization. He was asked to speak at our MSUD Symposium in Pennsylvania in 1988. We were amazed and thrilled that a man of his caliber would bring his wife and attend all three days of the meeting. We knew then beyond a doubt that his motivation was not a desire for personal honor and acclaim but he cared.

Dr. Guthrie once told me he uses the example of the successful organizing of our small group, to encourage others with disorders to do the same. His influence on our family and interest in our support group, prompted us to take time to attend the dinner. We appreciated the opportunity to speak briefly with Dr. Guthrie and his wife. It was a great honor to share this event with them.

After the dinner, several persons spoke about Dr. Guthrie's work and contributions in the medical field. One doctor spoke of the Guthrie test as one of the five major medical events of this century. A daughter of the Guthries presented a candid view of life on the home front, and several colleagues and friends told of experiences they shared with Dr. Guthrie.

Dr. Guthrie now in his late 70s, remained seated when he spoke briefly to the group. However, I am happy to report that his memory must be quite keen because he greeted Wayne and me by our first names. May God bless him for his lifetime service to mankind.

FORMULA COMPANIES RESPOND

Details

Written by Joyce Brubacher

Published: 17 July 2009

The editor of the National PKU News, Virginia Schuett, asked the three companies marketing medical food in the US to respond to questions PKU parents frequently ask about formulas. She summarized the responses in an article entitled "Answers to Your Questions about PKU Medical Foods (Formulas)" printed in the fall, 1994 issue. Mead Johnson, Ross Laboratories, and SHS North America provide formulas for MSUD and PKU so most of the information in the article is relevant to both disorders. I added MSUD references (italicized in parenthesis) where applicable.

Why can't the formula be made to smell and taste better, when it is the cornerstone of PKU (MSUD) treatment?

All of the companies acknowledge that the smell and taste of the medical foods (formulas) are problems for some people. All three have made major efforts in the past several years to improve the taste and smell, but this is not an easy task. The problem lies in the basic raw materials that need to be used. Formulas such as Lofenalac that are made to be low in phenylalanine through a chemical hydrolyzing process have a very distinct odor and flavor. Phe-free formulas (all of the other formulas distributed in the U.S.) are made of individual amino acids. Some of the essential amino acids contain sulfur and have a bad taste and smell. Certain non-essential amino acids required for extra nitrogen contribute to the undesirable properties. The vitamins and minerals that need to be included also add a strong taste. Due to the nutritional importance of most of the "offending" substances, it is not feasible to simply remove them.

Masking the flavor and smell is not easy either. No completely adequate coating to encapsulate the bad tasting amino acids has been found. Some flavoring agents actually strengthen the taste of the amino acids; some flavorings can trigger allergic reactions in some people. And of course not everyone likes the same flavor. Use of some ingredients that could improve taste would triple or quadruple the cost and they are not approved by the FDA.

One approach taken recently has been to decrease amounts of certain of the nonessential "bad-tasting" amino acids. The new formula distributed by SHS North America, Periflex, uses this approach. Phenex, (Ketonex) by Ross Laboratories, also is an attempt to improve the flavor and odor. Phenex (Ketonex) formula comes with an optional flavoring agent in two flavors. SHS is working on some innovative new presentations of the medical food that they hope will be available in the near future.

Why can't the formula be put into a pill form? Couldn't you just put in the necessary amino acids and leave out all the fat, carbohydrate, vitamins, and minerals to be obtained from another source?

This is a wonderful idea, but there are major obstacles to a pill becoming a successful alternative to other forms of the medical food. Even if only the required amount of amino acids were put into pill form, the pills either would need to be large, or the quantities required to replace all formula would be more than the average child or person could reasonably manage (40 to 60 or more pills per day!) Also, traditional pills require the use of binders and other ingredients to hold the tablets together. These ingredients cause tablets to be less soluble in the gastrointestinal tract. This would decrease the amount of amino acids available. Capsules could be used, but the shells contain protein in the form of gelatin.

There are other potential problems:

- If the medical food did not contain minerals and vitamins, there would be the risk of the child or young person forgetting to take the supplement and becoming severely deficient.
- For infant products, the Food and Drug Administration requires fat, carbohydrate, vitamins and minerals unless the company provides a major medial justification for an exemption.
- Unless a phe-free (BCAA-free) supplement of fat and carbohydrate is used to supply calories, persons on the diet cannot eat enough regular table foods to meet energy needs without exceeding their phe (leucine) tolerance.

Despite the problems, the Milupa Company of Germany experimentally produced a pill many years ago that was tried mainly in Europe. The pills were not well accepted in the quantities required and the idea was abandoned. Recently, one young woman during her pregnancy hand-stuffed one of the formulas into empty capsules in order to maintain the diet. But she had to consume 90 to 100 capsules per day and the capsules added 100 mg phe.

Still, the idea of a pill continues to have appeal. Despite the technical and other difficulties mentioned above, SHS North America is exploring a pill form of the medical food as one partial alternative. Clinical trials have taken place in Europe. Results of their marketing research and the regulatory environment here in the U.S. will determine suitability of this product for the North American market.

Why are the formulas so expensive?
As you are aware, the PKU (MSUD) medical foods are made of many compounds. These are obtained from raw material manufacturers located in various parts of the world. The basic nature of the formulas is very complex. The raw materials are rare compounds isolated from batches of starter material. They are extracted and purified to high food or pharmacological grade compounds. Amino acids, and protein hydrolysates treated to remove most of the phenylalanine (leucine), are simply much more expensive per gram of protein than is whole protein in normal infant formulas

Strict quality controls and laboratory confirmation of the manufacturing process also add to costs. Frequent analysis of the formulas is needed to insure that the ingredients meet label specifications.

These and all other costs associated with production are increased on a per unit basis due to the small production of the formulas. Regular infant formula is made in huge batches (60,000 to 120,000 pounds or more); so the cost of analytical tests required by the FDA for it prior to product release and during shelf life is proportionately much smaller per unit.

Also, because of the number of people needing the medical foods, batch sizes made at any one time must be small enough so that it is not outdated by the time it reaches the consumer. This increases costs per unit. In this regard, many state health agencies do not appropriately manage their inventory and often return products after their maximum shelf-life has passed. These products must be destroyed. At least two of the companies have liberal refund policies for outdated products, adding to costs.

Finally, mark-up on the price of the products when they are distributed through pharmacies can be very substantial and is not controlled by the companies.

The suppliers claim there is so much expense involved in producing the formulas for so few people that they don't make a profit on them. So why do we have so many different companies coming up with new formulas?
In the U.S., only three companies provide medical foods. They are dedicated to providing quality nutritional products for people with inborn errors of metabolism, not to making big profits. The

companies also help support many PKU-related activities such as parent and professional meetings. Mead Johnson, the first company to supply such products, has done so for over 35 years. A reflection of their interest in families is their program called "Helping Hands for Special Kids," which makes sure that children of financially needy families get the Mead Johnson formulas required (interested families should contact their physicians). Ross Laboratories entered the inborn errors of metabolism market a few years ago.

Major formula companies want to provide a complete line of products even if some products are made available at a loss to the company, for humanitarian or public relations reasons. This is also advantageous for selling products to some hospitals, and state health agencies that will not contract with a company for the regular baby formulas unless they have a complete line of metabolic formulas. SHS North America, a company based in England, is the only one of the companies that does not manufacture or market regular infant formulas. Their sole function is to provide disease-specific nutritional products that will help patients with PKU and other metabolic diseases.

"Three companies are better than one," I say. I think that we should all be glad to have the three trying to better the lives of those who have inborn errors of metabolism like PKU (MSUD).

Why couldn't there be a ready-to-use formula? It is very inconvenient to have to mix it up every day, especially when traveling or away from home.
There are many reasons why medical foods have been produced in powder form. These reasons include the following:

- Powders have a longer shelf-life than liquids (about twice as long). Because of shorter shelf-life, a liquid form of the formula would cost considerably more per gram of protein than powder (up to twice as much or more).
- Ready-to-use medical foods are difficult to make. This is due to the chemical reaction of amino acids and carbohydrate during processing and over the shelf-life of the product. Such a product would require careful animal and clinical testing to make sure all nutrients are available.
- Powders allow for easier individualization and modification to suit changing nutritional needs over a lifetime. Use of a powdered product also allows people to concentrate the formula so that needed protein and calories can be provided in a smaller volume.
- The processing required to make a shelf-stable, ready-to-use liquid would change the nutrient profile of the product and could alter the taste in an undesirable way.

Despite the potential barriers, SHS North America is exploring the technical aspects of producing a ready-to-use medical food.

TEACHING DIET TO YOUR CHILD: A DEVELOPMENTAL APPROACH

Details

Written by Joyce Brubacher

Published: 17 July 2009

This is a summary of a talk given by Cristine M. Trahms, M.S., R.D. at the MSUD Symposium in June of '94. Cristine is with the Department of Pediatrics, Division of Pediatric Genetics, University of Washington, Seattle, Washington. Several parents requested copies of the information she presented, and she graciously submitted this article. She informed me in February that a similar summary would appear in the proceedings of the 8th Annual Meeting of the European Society for PKU (ESPKU) under the title: Self-Management Skills: the Key to Successful Treatment.

Parents whose children have MSUD have been presented a challenge unique to parenting. Their tasks are to:1) provide metabolic balance for the normalization of growth and development and 2) support appropriate developmental stages of eating and relationship to food. The second task is more achievable if a framework for supporting developmental milestones is constructed.

Compliance with dietary management guidelines is the cornerstone of effective treatment for MSUD. The goals for successful self-management of MSUD, as we have defined them in our clinic, are that the child: 1) understands the basics of the disorder; 2) understands the food pattern and can identify acceptable and unacceptable food choices; 3) develops skills for making appropriate food choices; and 4) increases independence by learning how to prepare food, plan menus, and keep food records.

Parents are the initial managers of treatment. However, children with MSUD must themselves learn to adequately follow the therapy to the best of their abilities. To support children intellectually and emotionally as they begin self-management requires a basic framework built upon principles of learning. The method that has worked most effectively for our group is based on the principles of Piaget which have been widely described and have been used in many educational settings. The learning theories of Piaget described simply are: 1) that a child progresses through a series of stages of mental development and maturation; 2) a framework of readiness for intellectual development is necessary to promote learning; and 3) activity and exploration on the part of the learner is essential. We know that the progress of development proceeds in an organized fashion; that the child has an inner drive to make progress, but at an individual pace; and that our job as parents and professionals is to understand these developmental stages and support the child's readiness to move forward.

Piaget's model describes the developmental learning stages in a manner that outlines the capabilities of the learner:

1. The Sensory-Motor Stage-infants to age 2, who manipulate objects, are action oriented, are aware of present time, have representational thought and respond to their environment based on their own experience.
2. The Pre-operational Stage also called the Intuitive Stage-children ages 2-6, who have language and begin to respond to their environment based on semi-logical thinking, who are perceptively impulsive and blend fantasy and reality.
3. The Concrete-operational Stage also called the Practical Stage-children ages 7-11, who are eager to develop and use their skills at thinking, mathematics and reasoning.
4. The Formal-operational Stage also called the Reflective Stage-ages 12-15, which is adult style thinking with the ability to deduce and reason and learn to make decisions independently.

The practical application of Piaget's constructs is that:

1. Learning is based on cognitive readiness.
2. Parents and children must work together through the process one-step-at-a-time.
3. Parents work together to decide priorities for their child, that is, negotiable vs. nonnegotiable behaviors and what level of compliance is expected.
4. The more integrated the child's life pattern and the restricted food pattern become, the more likely long-term compliance will be achieved, that is, the food pattern is woven into family life rather than each food-related event individually negotiated.

These stages of learning can be translated into developmental self-management tasks for individuals with MSUD based on age and competence. The operational aspects of learning become more sophisticated as the child becomes cognitively more mature, but the goals themselves do not change. To be effective in promoting learning, education must be directed at the level of the learner. Additional information is added only when the learner is ready.

With the added stresses of management of a metabolic disorder, we must still remember that the development of a healthy relationship with food starts in early infancy with the response of the caregiver to the infant's cues of hunger and satisfaction. Some of these guidelines are:

- The food patterns of young children are shaped by many factors that parents can facilitate or ignore.
- Food acceptances of infants and young children are shaped by an innate preference for sweet tastes.
- The attitudes and acceptance of foods by adults and peers has a profound influence on food acceptance by the older infant and young child.
- Young children respond by accepting or rejecting foods based on the social context of their being offered-that is, a positive interaction from family or adults

offering the foods enhances acceptance and a negative or neutral interaction tends to decrease the probability of acceptance of a food.
- Acceptance of foods is also based on learning-that is, familiarity and presentation of the food in a form that is easy for the child to manage.
- Children learn from family and other adults how to regulate their own food intake-that is, they learn cues of satiation based on the social and cultural environment during meals.

These concepts can be translated into action by: introducing new flavors and textures gradually; enlarging the child's experience with as many forms of individual "yes" foods as possible; offering individual foods rather than mixtures so that foods may be appreciated for their flavors and textures; being patient with first efforts and allowing the infant to learn to feed himself/herself.

Table I (below) indicates tasks for young children. Younger school-aged children learn much from involvement in the process of food preparation. Cooking can be used to: 1) enhance a sense of accomplishment for the child; 2) have fun while increasing the child's self-esteem and self confidence; 3) support cognitive and social learning; 4) learn about the role of food as nourishment; 5) learn proper use of kitchen tools/utensils; 6) learn to weigh and measure foods; 7) learn to follow directions; 8) learn how to plan, organize and complete a project; 9) learn to work with other children/adults. These skills, when learned in early childhood, support effective self-management throughout life.

The tasks of primary grade children are shown in Table II (below). Children of these ages learn from problem-solving discussions and role-playing to practice the decisions that they need to make for themselves.

Adolescent children are responsible for the development of: 1) judgment; 2) appropriate responses to social pressures; 3) positive coping behaviors; 4) positive self-concept; and 5) assertiveness skills. The learning tasks of older children are shown in Table III (below).

In summary, we can expect children with MSUD to grow and develop at the maximum level allowed by their disorder by maintaining their therapy and having access to a learning environment that supports cognitive development and directs the development of appropriate self-management skills. A model for supporting the development of self-management skills is presented here.

References:

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Table I

Self-management Tasks for Young Children

Age (year)	**School level**	**Learning tasks**
2-3	Preschool	Learns to distinguish yes/no foods
3-4	Preschool	Learns to count foods: how many Learns concept of formula first
4-5	Preschool	Begins use of scale for measuring: how much
5-6	Kindergarten	Begins to prepare own formula with supervision Begins weighing foods regularly on a scale with supervision
6-7	Grades 1-2	Begins to list foods on food record

Table II

Self-management Tasks for Children

Age (year)	**School level**	**Learning tasks**
7-8	Grade 2-3	Prepares formula with supervision Lists food on food record Understands portion sizes
8-9	Grade 3-4	Prepares formula daily with little supervision Packs school lunch Chooses after school snack Prepares simple breakfasts Independently lists quantities of foods on food record

10-11	Grade 5-6	Prepares formula independently each day Prepares week-day breakfasts Prepares simple entrees independently Consumes full amount of formula independently each day

Table III

Management Tasks for Adolescents and Young Adults

Age (year)	**School level**	**Learning tasks**
12-14	Grade 7-9	Begins to independently manage total intake for the day Responsible for menu planning Responsible for food records
15-17	High school	Responsible for all aspects of self-management with continued parent support
18+	Post-high school	Transitions to adult based clinic care and independent living

NOTE: An Interesting Article by Dr. Morton

The Dec. '94, Vol. 94, No. 6 issue of Pediatrics printed a speech by Dr. Holmes Morton as a Special Article. In a note prefacing the article, the editor praises Dr. Morton's talk. He describes it as moving and thought-provoking-not your usual journal article.

Dr. Morton gave this speech at the 125th Year Celebration of Children's Hospital of Boston. It reveals the motivating force and factors behind the atypical doctor who established the unique clinic at Strasburg, Pennsylvania. It is a practical, earthy view of his work among common people and his dedication to it.

The article is too lengthy to reprint in our Newsletter. Dr. Morton has kindly given us permission to make copies which are available from our contact person, Dawn Marie Hahn. It is stimulating reading for anyone interested in metabolic diseases and not too technical for parents. Don't miss it.