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A NEW LIVER FOR KATHRYN

Details

Written by Mabel Burkholder

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The Norman Burkholder family lives about a 2 hour drive northeast of Lancaster, Pennsylvania. The Burkholders are an Old Order Mennonite family with eight children. Their daughters, Kathryn and Ellamae were born with MSUD. Kathryn was born in 1988 and Ellamae in 1992. Mabel, their mother, heard that some of the children with MSUD had bad teeth. Their local preventive care doctor suggested giving supplements of vitamin A to maintain healthy teeth and improve Kathryn's immunity.

Over the next several years, Mabel gave Kathryn vitamin A in doses she assumed were safe. However, vitamin A in excess is toxic. When Kathryn was around 5 or 6, she had toxoplasmosis twice which seemed to have caused her spleen to enlarge. Her liver was also enlarged at times. Her metabolic doctor, Dr. Holmes Morton, was concerned and ran various tests.

Tests showed a low level of vitamin A in her blood, so vitamin A toxicity was not identified until early in 1996 when Kathryn was hospitalized for five days. She was in the end stage of liver failure. The family now faced the issue of a liver transplant. Before the family made a decision, the doctor learned that the low blood levels of vitamin A were masking a vitamin A toxicity. Knowing the cause, they hoped that the liver could recover from the toxicity.

I had hoped to have a medical account of Kathryn's liver failure and transplant to print with this article, but it was not available. However, the transplant will be covered at the upcoming Symposium '98. Phone conversations with Mabel provided some of the preceding information.

On January 2, 1997 we were on our way to the Clinic for Special Children for a check up with Dr. Morton when Kathryn vomited blood. We didn't recognize the brown "coffee grounds" as old blood, but our driver did. Dr. Morton checked her leucine levels and then sent us to the Lancaster General Hospital. Our daughter Kathryn was in acute liver failure. Dr. Morton fought for her life for several days.

She was bleeding from a ruptured vein in her esophagus. They gave medication to quickly rid her intestines of blood because the blood was being digested. According to Dr. Morton, the leucine content of the blood was about equal to the amount of leucine ingested when eating 3/4 of a hamburger, which caused her leucine levels to reach 24 mg/dl. She was dizzy, groggy-like, with her eyes only half open the first day or two. To stop the bleeding that first night, they put ice water in Kathryn's stomach through an NG tube and drew it out again to help clot the blood. It didn't work. So they took her to surgery and cauterized the ruptured vein in her esophagus. The anesthesia made her groggy longer than normal. The anesthesia along with her high leucine level made it hard for her failing liver to work off all the toxins.

She wasn't allowed to have any "free" water by mouth that first night or the following day for fear it would encourage brain swelling. She was given lots of saline in her IV which made her beg for water. Dr. Morton thought he might lose her that night.

The second day, she still did not seem like herself. I thought her actions indicated brain damage, and I decided then we'll let her go and not do a transplant. That evening when Dr. Morton came in, he told me I couldn't judge by what I was observing. Her poor responses were due to the toxins still in her system. It was taking longer to work off the toxins because of her failing liver. Her leucine was no longer elevated and was not affecting her. So the whole turmoil of indecision about a liver transplant started up again!

The half year before this episode - after discovering her liver failure - Dr. Morton was trying to balance the amino acids to keep her MSUD under control. He did not give a bit more than necessary, so her liver wouldn't have to work with an excess of amino acids. Two companies supplied individual amino acids which Dr. Morton used to make a formula. He used Farm Rich, a non-dairy creamer, and the mix of amino acids he thought best. He tried to liminate all Vitamin A from her diet in order to reverse the liver damage.

After this big bleed, Dr. Morton was afraid Kathryn might not make it much longer. He wanted us to decide whether we're going to do a liver transplant. When we went home that night, I explained the situation to the family. There is no chance of life as she is now, and she would have a 65% chance with a liver transplant. We asked our children what they think we should do. Melinda said, "Oh, give her a chance." And Edwin, the one who often fought with Kathryn, said, "Yes, do the surgery!"

Well, we felt lost, and you always think doctors may be making it sound worse than it is. We didn't decide right away, and then she "leveled out" a bit - stabilized. The third day, she actually understood her schoolwork again.

After we were convinced the doctors were right about her chances, we said okay to the transplant. But by the time we'd made a decision, she was so much better and no longer an intensive care patient. She wasn't eligible for the number one category anymore. If we had made our decision the first night, she might have had a new liver in 24 hours, Dr. Morton told us. With the decision to do surgery, I prayed that God would take her before the surgery if the new liver was not going to be a good match, or if we should not do the transplant at all. The way Dr. Morton talked it seemed her life was very much "on the edge."

We realized later if we would have decided to let her die, her stay in the hospital might have been even longer. She couldn't eat because of the pancreatitis and Dr. Morton didn't think she would have been able to eat again.

We are also glad we didn't have to do the transplant then. We have heard since of other persons who were given a mismatched liver when it was a life-and-death matter and later needed another transplant.

During the month and a half that Kathryn was in the hospital before the transplant, she developed pancreatitis, so she had to stay on an IV the whole time. The back pressure caused by the blocked liver created pressure on her pancreas and lungs.
Kathryn had low oxygen saturation for a year or so before this episode. Pressure built up from the blood not being able to go through her liver freely and created shuntings in the lungs. The blood actually bypassed some of the oxygen-making places in the lungs, going through the little shuntings created by the extra pressure. That same pressure caused varicosities (varicose veins) the thickness of an index finger all the way down her esophagus into her stomach. Dr. Morton had been afraid of having those veins rupture during the half year that we tried to reverse the toxicity of the liver but I'd forgotten he said that.

She was coughing so much the last week before her big bleed, coughing hard every time she drank her formula. She wanted it quite warm and asked for "quick water" (water quickly) after her formula to keep her cough from making her throw up. We realized later anything cold probably aggravated those big veins in her throat dreadfully. But since I'd forgotten about the possibility of varicose veins, I didn't tell the doctor about this reaction right away.

Being on IV's for a month made her liver worse, so her pancreas didn't get better either. Her eyes got very yellow in the corners. Some days she was better and we almost had hopes of bringing her home.

I'm glad now that I didn't know that Dr. Morton believed she would never be able to eat food again without having a new liver. Knowing this, and that being on the number two list for a liver transplant meant it could take half a year before a liver was available, would have been quite depressing. I believed she would get better and could go home.

The third week in January she was taken to Philadelphia by ambulance (without sirens) to have an evaluation. After one week at Philadelphia, she was back at the Lancaster Hospital which felt like home by now.

During this time Kathryn was on IVs which advanced the liver failure, and Dr. Morton kept fearing another bleed. This would cause very serious problems with her MSUD. He tried to get the officials from the transplant center to hasten the transplant. The officials finally said they would discuss metabolic patients getting priority status at their next meeting.

Before two weeks were up, Kathryn had another bleed. It was not as scary or as big a bleed as the first time, but, by needing intensive care, it put her back on the number one list for eligibility.

So we rushed Kathryn to Philly, sirens going this time, and admitted her to ICU in the Children's Hospital of Philadelphia (CHOP). To me it was "just a comfortable little bleed," even though she did pass out from toxins, etc.

As we entered CHOP, I was scared to be without Dr. Morton for fear Philly doctors would miss something with her complicated case. After being there almost a week, they found a good liver.

The first liver they'd prepared for her was turned down by Dr. Shaked, which increased my faith in him. He knew that many doctors, nurses, etc. had been working towards a transplant, and yet he was able to refuse the liver because it had abnormal routings, and Kathryn was stable enough at the time. He could've used it if she'd been too sick. However, the extra rerouting of tubes to connect that liver's abnormal routings to Kathryn's could have caused more chance of rejection. We've been relieved often thinking about it. She barely had <u>any</u> rejection with the liver they found for her three days later.

The week before her transplant, while Kathryn was in the ICU, she got so much better that Dr. Morton was afraid she'd be taken out of ICU and put on the regular floor. That would put her on number two status again. Then he would've had to struggle to get her through the next big bleed. We believe God saw to it that there were little problems that week in ICU - enough to keep her in the ICU and on the number one list for a liver. Kathryn's insulin and glucose levels fluctuated, and another bleed one night caused her leucine levels to elevate to 14 mg/dl again. When I got uneasy about something, I'd call Dr. Morton.

Kathryn's liver came from a small 22 year-old woman on Feb. 12. They had prepared to give an adult in an adjoining hospital the right lobe of the liver and Kathryn the left (smaller lobe). However, the liver was small enough to fit the complete liver into the cavity from which Kathryn's failed liver was taken.

Before the hallelujah of coming home, we had many ups and downs in the ICU and later on the seventh floor. For five whole weeks we experienced both anxiety and hope after surgery. Reading back over the diary I kept, it now looks like this was the biggest cliff we plunged off yet.

When we had questioned Dr. Morton about the shuntings and the low saturation problem, he said it would reverse itself immediately after the liver transplant. But Kathryn's oxygen saturation remained low after the transplant. Dr. Morton's explanation made sense to me - when they used hyperalimentation *(TPN by IV)* after the surgery, the new liver was filled, making it larger which caused slight pressure on her lungs, enough to keep the shuntings in her lungs open.

His theory proved correct. When IV intake was decreased in preparation for going home, her oxygen saturation jumped from 60 to 80%. In a week and a half after being released from the hospital, they tested 99%. <u>Relief</u>! We thought that problem would vanish immediately after surgery, but it took a month! Now no more purple lips and fingernails and a normal flesh-colored face for Kathryn, just like other children! No more getting short of breath from walking or playing.

Kathryn was discharged from the hospital on March 21. After Kathryn was at home, she started eating better. She'd been unable to eat for one and one-half months before the surgery - maybe that's why it took awhile for her appetite to return. Or maybe because of so many medicines. Anyway, she'd started eating ham roll and other meats quite heartily about one month before her two months of tummy aches started.

After she started having tummy aches, Dr. Morton thought it might be an ulcer, so we tried to get scheduled for an endoscopy. I guess other parents would've said, "Here we come, she needs to be hospitalized." But we struggled to feed her for four weeks until she finally got scheduled for an appointment.

By that time, I was afraid she was dehydrating from throwing up several times a day. She would lie on the sofa with tummy ache for hours at a time, sometimes throwing up tiny clots of blood. I was afraid her stomach would rupture and hemorrhage if we let it go one more weekend! Finally they checked her at CHOP and kept her.

The doctors diagnosed an ulcer with lymphoma around the ulcer - a drug-induced lymphoma they said. So she was given acyclovir by IV. In Philadelphia, they said if they can't heal the lymphoma by withholding the Prograf, they would use chemotherapy. (Prograf is an immunosuppressant drug to prevent the body from rejecting the liver.)
In the meantime, I was in contact with friends who had been treated in a hospital in Pittsburgh. The doctors in Pittsburgh were much more experienced with lymphoma. Philadelphia had 18 cases, but Pittsburgh had probably a hundred or more. Pittsburgh treated all their cases without chemotherapy.

We were at CHOP for 2 , weeks (Aug. 12 to 28, 1997) and at home with an oral acyclovir for the lymphoma for 1, weeks. To heal the lymphoma, they had to withhold the Prograf for as long as the liver didn't show signs of rejection. When Kathryn was released from CHOP, she tested positive for the Epstein-Barr virus, so they changed the oral acyclovir dosage to a therapeutic level every 8 hours plus one dose at nighttime. Kathryn went to the first week of school with some tummy aches.

Dr. Reyes from Pittsburgh said their research shows that oral acyclovir is worth almost nothing in treating the Epstein-Barr virus. It seemed to prove him right when by the end of the week at home, Kathryn's tummy aches made her cry once or twice a day again. By then we had scheduled a trip to Pittsburgh to give her a thorough check up and get a second opinion.

Well, the second opinion turned into another 2 , weeks stay at the Pittsburgh hospital (Sept. 8 to 25, 1997). But she was the healthiest child on the floor and to her it was a "joy ride." We felt like the luckiest parents on the floor. We also came to realize that no matter what hospital you are in, Epstein-Barr virus and lymphoma in a transplant patient are very hard to treat correctly.
One boy at Pittsburgh had the same lymphoma as Kathryn. He had been treated in Boston first with chemotherapy which actually made the lumps grow. We felt heaps of relief that we went to Pittsburgh before Kathryn was given chemotherapy. I sincerely thank the three hospital staffs for doing all they could for Kathryn and us.

Two days after Kathryn was admitted to the Pittsburgh Children's Hospital, they replaced the oral ayclovir with IV gancyclovir. On her fourth day there, they put a scope down into her stomach and discovered CMV, a virus that isn't touched by acyclovir. Can you imagine the relief we felt that it was discovered! We were very glad we had sought a second opinion. Now we were confident that putting her in the hospital, even though she seemed much better, was the right move.

We had been afraid the doctors would miss just one little thing that would make all the difference. Pittsburgh had a great deal of experience with liver transplant follow-up and we liked the way they treated aggressively.

We also heard of a child that was given only one lobe of liver and later got leakage of either blood or bile causing infection in the intestinal cavity. That makes us glad that God directed it all and Kathryn got a whole liver (which is not the usual).

Right now Kathryn's at home and very healthy. Just Friday morning, it amazed me again that she can actually jump on her bike and easily make it up the little hill past our barn. For the last few years, others had to use a wagon or cart to take her out the lane to the school bus stop. She's actually chubby now (without any Ensure or IV) and back to liking some meats. While she had tummy aches, she preferred all low protein foods - because protein is harder on a sick tummy? She still likes MSUD formula on her cornflakes and likes Cremora "cheese" sandwiches made with rusks, or rusks buttered and browned on both sides. She prefers low-pro bread to ours (even though the last loaf I made is coarse and hard) and sometimes helps Ellamae eat her low protein noodles or soup. Tomato soup is still better her way (Cremora instead of milk). But I have to think, which one of us could change from eating our cornflakes with milk to eating them with MSUD formula! It is the same for her - it doesn't taste right with our milk.

We want to thank the many people who supported us with cards, letters, phone visits, prayers and financial support. All of these helped us get through this ordeal. Looking back, it looks almost impossible to have stood it all without completely collapsing. Our thanks to everybody, especially God.

UPDATE:

Kathryn missed a lot of school and was so miserable before her transplant. Now she loves school and her learning is much improved. She is nine and repeating the first grade. Because of all the attention she received, she does have a problem with temper tantrums which the family is dealing with.
Kathryn enjoys her baby brother, born on October 29, 1997. He has MSUD, so the family still has two children on the MSUD diet, baby Norman Jr. and Ellamae, 5. Kathryn is gradually giving up her interest in her old diet and adjusting to being able to eat any foods. However, she still likes to eat some low protein foods made for her brother and sister and drink a little formula when she gets a chance.

Baby Norman has been a healthy baby and weighs 17 lbs. at 4, months. He was tested for MSUD within 24 hours of birth and started on the diet on the second day of life. He has had only one

hospitalization - an overnight stay at the Lancaster General Hospital. At that time he had an asthma-like tightness that was treated with a nebulizer. It seemed he got tight from the slightest breeze or a cold room, but only when his leucine was too low. He gained weight so fast the first three months that his leucine level was often way too low. His mother started sending blood on filter paper to Dr. Morton by overnight Fed-Ex twice weekly at times. This helped.

Although Kathryn is doing really well, a liver transplant is not the cure-all for MSUD. It is a risky procedure and very expensive. The cost for Kathryn's family, who has no medical insurance, was over $500,000. Costly anti-rejection medicine and follow-up treatment will continue throughout Kathryn's life. After Kathryn's transplant, the local community pitched in with fund-raisers. One auction raised $27,635 for Kathryn's medical fund.
This family had many exhausting challenges in the last couple of years but through their experiences, many persons have become aware of MSUD. Medical research has been advanced. Kathryn is enjoying life. The Lord has brought blessing out of adversity.

IMPROVING PKU (MSUD) DIET COMPLIANCE

Details

Written by Teresia Goldberg

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By Teresia Goldberg, MS, RD and David Pelcovitz, PhD.

The following article is reprinted with permission from the *National PKU News*, Vol. 7, No. 3., Winter 1996. Although written specifically for the PKU population, the article has much good advice for those dealing with MSUD diets. Just think MSUD instead of PKU.

Ask any parent what the greatest challenge of the PKU diet is and the likely answer will be "the medical food." Some children may begin to refuse the "milk" from an early age. The problem may last only a few days, or can go on for a long time. The struggle that begins can negatively affect relationships between parents and other family members as well as the child's metabolic control. To help families with this and other compliance problems, we invited Dr. David Pelcovitz, Chief of Child and Adolescent Psychology at Cornell University Medical College to speak to our Metabolic Center parent support group at North Shore University Hospital in Manhasset, New York. We later put the ideas into a paper from which this article is drawn.

(Pelcovitz, D., and Goldberg, T., Enhancing nutrition compliance in children: Inborn errors of metabolism as a paradigm. *Topics in Clinical Nutrition,* 10 (2): 73-81, 1995.)

To prevent power struggles over feeding, parents should have an attitude of calm control. But this is not easy when managing children with PKU. All parents know that not following the diet can lead to serious consequences. This makes it difficult for parents and other care givers to take a "low-key" approach when the medical food or other foods are refused. Also, most problems related to poor compliance are not seen immediately, adding to the difficulty. For children, who by nature think in concrete terms, the lack of immediate feedback is especially difficult. In this article, we will make specific recommendations to help diet compliance.

When the child begins to eat table foods, the diets of children with PKU noticeably differ from those found in normal meal patterns. Not only must they drink adequate amounts of the medical food, but they must avoid eating many of the foods eaten by family and friends. Although there are a variety of modified foods, their appearance and taste can differ somewhat from those of their regular counterparts.

The potential for noncompliance begins when children reach an age where they start to have a say in what they eat. It is very normal for toddlers to get into power struggles. At this age, almost all parents report significant problems in properly managing their child's diet. In a European study of eleven children with PKU, parents ranked the diet as their most difficult problem. At our metabolic center, eight out of ten families of toddlers report becoming involved in intense struggles with their children over drinking the medical food. Feeding problems in these children have been reported as early as eighteen months of age.

Children of that age typically are dealing with their beginning independence. It is not surprising that the child with PKU may now begin to refuse to drink the medical food. Families may use various techniques to help this problem, including concentrating the medical food, flavoring it, using reward systems, etc. Battles may continue for days or weeks at a time, occasionally ending in forced feeding or noncompliance. At this age, there is also increased exposure to table foods, providing more opportunities for power struggles over what and how much is eaten.

Don't be overindulgent

Parents may have strong emotions due to having a child with a metabolic disorder. These emotions may include feelings of guilt or pity because of diet restrictions. The feelings may result in parents having difficulty setting limits. Remember that the youngster you are dealing with is a child first, and a "patient" second! The scientific literature on children with chronic conditions describes the danger of responding to them by becoming more indulgent, or by placing fewer demands on them. A cycle may be set in motion. The child wants to gain reassurance that he or she is like everybody else and tries to get care givers to "normalize" disciplinary practices. This would prove the child is no different from siblings or friends.

For the child with PKU, eating is often the arena where such battles are fought. As a parent, you may need help getting in touch with underlying feelings of guilt and self-blame and in finding positive way of dealing with them. Do not apologize for the restrictions to your child

or to others in the presence of your child. Recognize that consistent regulation of your child's diet is *an act of love,* even in the face of tears and angry rejections.

A related problem is the tendency for some parents and care givers to encourage unusually high levels of dependency in the child. Concerns related to possible consequences of having PKU often result in overprotectiveness. Children with PKU have been shown to be more dependent than non-PKU adolescents. Although the high levels of concern are understandable, these feelings may set the stage for heightened rebellion. Conflicts over diet limits are a likely battleground in the struggle of the overprotected child to reach independence. The important thing is not to "baby" your child. Toddlers should be weaned to a cup, be expected to self-feed and be treated at the table in a way that is appropriate for the child's age.

Avoid power struggles

Recent research, including children with diabetes, shows that when parents display a high level of expressed emotion there is more likely to be poor diet compliance. Examples of "expressed emotion" are criticism, overprotectiveness and intrusiveness. But when care givers dealing with the children are helped to become more calm, consistent and supportive, the situation improves. Unfortunately, having a child who can have serious problems because of diet noncompliance is a situation that can easily lead to high levels of expressed emotion. For healthy children, guidelines for eating are straightforward. Power struggles are avoided by care givers taking responsibility for when and what the child eats, *but the child taking responsibility for how much and even whether he or she eats.* Care givers of young children with PKU cannot afford the luxury of their children deciding the quantity or even the timing of the food they consume.

Recommendations for Improving Diet Compliance

Issue	Associated Problems	Recommendations
Guilt Tendency to foster dependence in "special child" Power struggles Coordination of care givers	Difficulty setting limits Overindulgence Overprotection Child views eating as battleground Child receives inconsistent messages	As a parent or care giver, try to view limits as necessary and not as punishment. Offer allowed foods only. Redirect: plan distractions ahead of time, redirect your child's attention to favorite non-eating activities such as games or books (for example, "This might be a good time for us to read," or "How about going out to ride your bike?" Be assured that without pressure your child can make the right decisions.

		Learn appropriate expectations for the child's age from your clinic. Allow your child to participate in measuring the quantity of allowed food by no later than age three years (for example, "Let's choose your snack and count together how many pieces you will take.") Be aware of the wisdom of "pulling back" when faced with increasing conflict. Remember that young children use feeding as an arena for exercising control. Don't present eating as a request (for example, "Do mommy a favor and eat this"). Instead, firmly say, "It is now time for your drink." To de-escalate conflict for children under age three, briefly leave the room or hold the child calmly for several minutes without talking about eating. For a child over age three consider use of time out; offer choices for the medical food in flavor, consistency and temperature. Recognize the importance of parents working together as a team. If you are the parent who is detached or on the periphery of diet management, try to become more actively involved.

Child psychologists have found that when parents are in situations where they have little control, they are likely to deal with their children in a way that is characterized by high levels of emotion and criticism, and low levels of praise. But studies show children eat *less*, not more, when care takers become too active. Children quickly learn to manipulate their care givers' desperation to get them to eat. (Statements like "Do mommy a favor and drink this" are counterproductive.) Once the child realizes his parents are no longer emotionally invested in all his or her actions, cooperation will improve.

One very important thing that parents can do to reduce power struggles is to give the child the feeling that he or she has some control over a situation that is restrictive by nature. Whenever possible, parents should offer the child an element of choice. For example "You can either take your drink from the red mug or the blue cup." Also, children can be allowed

some control over preparation of their food and the medical food. For example, for the medical food, the child can be given a choice of flavorings (chocolate, strawberry, etc.), of consistency (how much water is added) and of temperature (room temperature, cold or even frozen).

Coordinate teamwork
It is not uncommon in families of children with chronic conditions for one parent to become over-involved with the child while the other takes a very minor role. Most often, mothers are intensely involved in the day-to-day care of their child while fathers flee into the world of work. More equal distribution of responsibility of care givers may be a crucial factor in improving compliance. Sometimes, a mother's over-involvement adds to the emotional intensity of the situation. Her expressed emotion becomes greater and power struggles increase. If one parent has the burden of exclusive responsibility for the diet, the level of stress is great for that parent. A study of families of children with PKU has shown that parental cooperation and family cohesiveness are important for dietary adherence. Often the parent who is less emotionally caught up in the child's diet may be the preferred parent to supervise the child's diet precisely because of the greater detachment.

Consider a reward system
Formal reward systems at times have their place in PKU management. Use of stickers and other reward systems can jump-start a stalled situation marked by a power struggle. However, care givers need to be careful that the child does not view such rewards as a subtle form of pressure. In many studies the children who were rewarded for trying new foods showed less enthusiasm for the food than children who were not rewarded. If after a short trial you find the use of rewards causes increased resistance or conflict, simply stop the rewards. If you or the clinic decides that a reward system is worth trying, it is important to make sure that the reward is age-appropriate, the chosen item or privilege is one that the child can get excited about, and it is realistic for the parent to give the reward quickly. Some examples of non-food rewards which have proved effective for preschool-age children include extra bedtime stories, stickers, puzzles and small toys. If a longer term reward is appropriate, a trip to the park or zoo could be used. Children ages six to twelve can be offered baseball cards, later bedtimes on a weekend, extra time alone with a parent, or a chance to attend a concert or sporting event. Consult with your PKU clinic if you have difficulty carrying out a reward system. The clinic should make recommendations based on family dynamics, or work with you and a mental health professional to carry out a plan to deal with the problem.

CHECKING OUT NEW FOODS

Details

Written by Elaina Jurecki

Published: 20 July 2009

By Elaina Jurecki, M.S., R.D., Kaiser Permanente Medical Center, Oakland, CA
Adapted with permission from the *Organic Acidemia Association Newsletter* (Vol. VI, No. 2., Aug. 1996).

So you are walking down the isle in the grocery store and wondering whether or not a new food you see on the shelf is okay to include in your child's diet. Now-a-days it seems like there is a new food item every time you go into the store. This is especially true with cereals and snack foods that can frequently contribute to a large portion of your child's intake. The best place to look to determine if this food is okay is by checking out the label and evaluating its nutrient content.

The food label was revised a couple of years ago and now includes a lot more useful information. The new label is illustrated here. This information is provided by the manufacturers and regulated by the U.S. Food and Drug Administration (FDA). The nutrient content is listed per serving size, and the number of servings per container is also indicated. Serving sizes have been standardized by the FDA to allow for comparing nutrient content of similar products made by different manufacturers.

You can now find the amount of calories per serving size and the number of calories that come from fat. The total amount of fat is listed in grams and includes the amount that comes from saturated sources. The cholesterol and sodium contents are listed in milligram amounts. The carbohydrate content provides the gram total amount and that amount from dietary fiber and sugar. The total grams of protein content is also listed.

The percent of Daily Value listed next to several of the nutrients is a new reference value. This is used to determine what the recommended intake per day should be based on a daily intake of 2,000 calories. The chart at the bottom of the label shows Daily Values for fat, saturated fat, carbohydrate, and fiber for the 2,000 calorie diet and for a 2,500 calorie diet. Consumers can use the Daily Value percentages to see how a food fits into an overall diet. It should be kept in mind, however, that Daily Values may vary, depending on an individual's calorie needs. Keep in mind that these calorie needs are more typical for an adult's intake, and for an individual not following a special diet.

The grams of protein, fat, and carbohydrate have been rounded to the nearest whole number. In other words, a food that contains from 0.0 to 0.4 grams will be listed to contain 0 grams, and a food that has 0.5 to 1.4 grams will be listed to contain 1.0 grams. Therefore, you may select a food that shows zero grams of protein, but it actually could contain up to 0.4 grams of protein per serving. This difference in protein content could cause an individual with a disorder in protein metabolism to get out of metabolic control and even result in hospitalization. So what should you do to verify the actual protein content? Contact the manufacturer for that food to find out the exact protein content. You can also verify the amounts of other nutrients listed. The name, address, and telephone number (and in many instances a toll free number is provided) for the manufacturer will be listed on the food product.

The ingredients of that food will also be listed on the label. The ingredients are listed by descending order by weight. For example, when the amount of each ingredient is weighed, the ingredient that weighs most is listed first. The ingredient that weighs the least is listed last. Ingredients that make up 2% or less of a food are listed at the end of the ingredients

listing in no particular order. Because of the fancy terminology for ingredients food companies use, you may need help with determining what they actually are. For example, cellulose gum is a fiber, corn syrup solids are simple sugar, and mono and diglycerides are fats. Check out these ingredients with your nutritionist to make sure that the food is acceptable on your child's diet.

The only vitamin and mineral values listed on food labels include vitamin A, vitamin C, calcium and iron and are indicated as a percent of Daily Value based on a 2,000 calorie diet. Additional information regarding the vitamin and mineral content of foods can be found in food composition tables. You may need to know the amounts of other nutrients that are not included on the label. For example, an individual with MSUD needs to know the amino acids: leucine, valine and isoleucine content for that food. A person with a disorder in a long chain fatty acid oxidation may need to know what the essential fat content is for that food. This information can be found in published food composition tables. There are also many computer nutrition programs available that include this information. The USDA Nutrient Database includes the largest amount of food composition data for over 5,000 different food items. This governmental agency has previously published their food composition tables, but now makes it more readily available via the Internet.

So don't be afraid to check out new foods. They may help to provide more variety in your child's diet. This is especially important when the diet is restricted in some way. But if you are unsure whether the food is truly acceptable in your child's diet, or if you are unsure of how much to give your child, then the safest thing to do is contact your nutritionist and/or Metabolic Health Care team for further assistance. Bring the food labels with you to your next appointment with your Metabolic Health Care Team, and they can help you determine if this is a good food and/or how much your child can eat.

DRINKING THE FORMULA ONCE, TWICE OR THREE TIMES A DAY: DOES IT MAKE A DIFFERENCE?

Details

Written by Dr. Miriam Keller

Published: 20 July 2009

This article appeared in the Spring/Summer 1996, Vol. 8, No. 1, issue of the *National PKU News*. The information is quite applicable to MSUD. The author, Dr. Miriam Keller is a young German physician who has PKU.

For many people it would be much easier to consume the entire amount required of the formula (medical food) in just one sitting. Particularly for teens and young adults who are

frequently away from home, drinking the formula is awkward and inconvenient during the day. Most families have been told, however, that it is ideal to divide the required amount into at least three portions. These portions should be consumed at intervals throughout the day. This recommendation is based on well- known principles of amino acid metabolism.

Several recent studies in Germany have confirmed the importance of consuming smaller amounts of formula at intervals, rather than in larger amounts once or twice daily. I want to summarize and explain the investigations of the University Clinic in Berlin. The team there, of which I was a part, involved a special work group [consisting] of a nutritionist, scientists and physicians dealing with the problem of PKU formulas.

Nitrogen is the most important building block of amino acids, and thus of body protein. The formula is the main source of nitrogen for persons with PKU *[and MSUD].* It provides amino acids (containing nitrogen) used for making body protein. For optimal protein synthesis, the body requires the presence of amino acids at intervals throughout the day, in combination with carbohydrates or fat. But amino acids cannot be stored, and any surplus that cannot be used at the time is broken down to be used for energy. To become a source of energy, the nitrogen from the amino acids is removed and excreted in the urine.

Several studies have shown this effect. In one study, ten young people with PKU (ages 12-26 years) ingested their formula on one day in a single or only two doses. Another day, they divided the amount into three portions. On both days, they collected their urine and the amount of nitrogen excreted was measured. The amount of nitrogen in the urine is an indicator of the quantity of amino acids that cannot be used for protein synthesis, and are therefore broken down and excreted. *The study showed that significantly more nitrogen was found in the urine when the formula was ingested only once a day.*

In another study, the same procedure was done for a 22 year-old female patient on a strict low phe *[phenylalanine]* diet. On one day, the formula was divided into three portions and ingested with the main meals. On the other day, the entire daily amount was taken during breakfast. Again, nitrogen loss increased when formula was ingested in one portion. The study also used a stable isotope as a marker to measure amino acid use. Amino acids are broken down into "carbon skeletons," and finally into carbon dioxide if they cannot be used for protein syntheses. The carbon is exhaled in breathing. In this study, the isotope was ingested along with the formula. As expected, the isotope elimination rate was higher when the formula was taken in only one portion. The larger the amount of amino acids consumed in a single dose, the greater the breakdown.

Both studies show the importance of consuming the formula *with food*, well-distributed during the day. Consuming the formula in only one or two doses results in poorer use of amino acids for protein synthesis. Also, the person who ingests the entire daily formula during a single meal misses the benefit of amino acid supplementation for the rest of the day.

Clinics should remind their patients of these findings. Families should try to take the advice to heart. And if you have PKU *[or MSUD]*, you should reflect on your own habits. Can you optimize your daily routine based on the findings of these studies? Do not hesitate to do the best you can for your health!

THE DIET AND SCHOOL

Details

Written by Sandy Bulcher

Published: 20 July 2009

Sandy Bulcher shares how their family has handled Jordan's diet at school. These good, useful ideas, are helping her son to become independent. The article is reprinted from the newsletter of the Association for Neuro-Metabolic Disorders (Vol. 17, No. 3, Dec. 1997). The newsletter is affiliated with the Pediatric Neurology Metabolic Clinic at the University of Michigan Medical Center.

I'm sure that we are no different than most parents. After being told that our son needed to follow a specific diet, we wondered: How is he going to follow a diet at school? What about parties at school? Will he make appropriate choices when we are not around? Should we send formula to school?

Jordan is in the 2nd grade now. We've had a few years to figure out how to manage his diet at school. Fortunately, the school staff has been very cooperative and eager to learn about his MSUD diet. In the evening, we review the next day's lunch menu. Jordan decides if there are enough low protein food choices to satisfy his appetite. If not, I pack his lunch. Jordan's favorite packed-lunch is a butter sandwich, chips, dill pickle, and a fruit rollup. His favorite school lunch is spaghetti (no sauce), garlic bread, salad, and applesauce. His leucine tolerance is 1000 mg/day. He brings a notebook to the cafeteria each day and the lunch aide records the kind and amount of food he eats. I decide what to give him for dinner based on the amount of leucine that he had at lunch.

Recently, the aide was absent, so Jordan himself recorded what he had eaten. When he came home from school, he announced that it was "easy" to keep track of his diet and that he would be recording it from now on. Needless to say, we are thrilled that he is being so responsible. Incidentally, I went to school during lunch time many times his first year, so that I could see what size portions were being served to determine the amount of leucine.

At the beginning of the school year, Jordan and I went shopping for his favorite "zero protein" foods to be used during parties. He chose fruit rollups, Mike and Ike candies, and low protein wafers. When one of the students brings in a birthday treat, his teacher gives Jordan a choice of one of his snacks and sends the birthday treat home in his lunch box for me to decide if it's appropriate. This has worked well and he doesn't mind not having the same snack as the other children. And I know that he is not getting any more leucine *[at school]* than what he had at lunch.

Jordan really likes the breadsticks they occasionally serve at lunch. In his notebook recently, it was recorded that he only ate half of the breadstick. When I asked if he wasn't

as hungry at lunch, he replied, "The breadsticks were different today, a lot bigger, so I only ate half of it so I would not get too much protein." It was refreshing to hear that he had thought this through and made a good choice. Clearly he is becoming more independent.

Figuring out how to handle his formula at school has been more challenging than his diet. Jordan does not want to drink his formula in front of anyone other than us at home. Along with his teacher, we worked out a system. He brings his "special drink" to school in a thermos in a small cooler. When he gets to school, he puts the cooler in a conference room next to his classroom. (He doesn't even want it in the same room.) He drinks juice at lunch time and drinks his formula when he comes in from recess. He said that he slips into the conference room while the other children are getting settled in and nobody sees him drink his formula. He's been happy with this arrangement.

Overall, it has been easier to manage Jordan's diet and formula at school than we expected. It is critical, however, to have a teacher who is flexible and supportive. Jordan is doing well in school and the majority of the time having MSUD is not an issue for him. **The temptation we all must check as parents is the urge to overprotect. There is a fine line between mothering and smothering and fathering and bothering.**

FEATURED FAMILIES - MSUD: FACT OR FICTION

Details

Written by Paula Ruter

Published: 20 July 2009

This account is reprinted from the newsletter of the Association for Neuro-Metabolic Disorders (Vol. 17, No. 3, Dec. 1997). The newsletter is affiliated with the Pediatric Neurology Metabolic Clinic at the Michigan Medical Center of which Dr. Richard Allen is the director and Cindy Parker and Anna Marie Schaefer are nutritionists.

My husband, Lance, and I have always been voracious readers, especially before we had a baby to occupy our time. One of our favorite authors is the mystery writer Patricia Cornwell. In her book, *Postmortem*, one of the characters has a very distinctive trait. His body odor has a sweet smell, like maple syrup. The author goes into great detail describing a condition called Maple Syrup Urine Disease. We thought that this showed great creativity, to "make up" such a strange disease. Little did we know what an impact this "figment of Ms. Cornwell's imagination" would have on our lives.

On July 17, 1997, I gave birth to our first child, a daughter we named Anna Augusta. While still in labor, I read the brochure on newborn screening. I was surprised to see MSUD on the list of diseases. I remember showing it to my husband and saying, "Look, this is actually a real disease!" We would soon find out just how real.

One week later we received the call from the pediatrician's office telling us that one of her newborn screening tests had come back abnormal. By this time Anna was exhibiting what I now know are the usual symptoms - irritability, high-pitched cry, refusing to nurse and arching her back. Also, looking back, her urine did have a syrupy, sweet smell. Of course, as a first time parent, I thought that everything about a baby was supposed to smell good, including her dirty diapers! The next day we were sent to the University of Michigan Medical Center, where we met Dr. Allen, and our long journey with MSUD began.

Though Anna's first three weeks were rocky, it has been almost smooth sailing since. She is now a happy four month old who smiles frequently and loves to be cuddled. Dr. Allen, Cindy and Anna Marie have been wonderful, along with our local pediatrician, Dr. Debra Simms. Although there are many ups and downs ahead, we feel up to the challenge. It still amazes me, however, that a work of fiction could become such a fact of life for us.

UPDATE: Anna continues to do really well at nine months and weighs 15 lbs. 10 oz.

FEATURED FAMILIES - OUR BABY'S STRUGGLE TO LIVE

Details

Written by John & Miriam Shirk

Published: 20 July 2009

John and Miriam Shirk had been blessed with five healthy children before Ammon was born. They are Old Order Mennonites living in Liberty, Kentucky. When reading the following story, keep in mind that they choose to have no phone or electricity. They do not drive cars, but use horse-drawn vehicles or ride with other people. Ammon's treatment was started in Kentucky when he was 17 days old and continued by Dr. Holmes Morton in Pennsylvania from the 19th day on. Details about the treatment of MSUD are of great interest to me and I presume of interest to others also. So I have included a few notes of information (in italics) that were supplied at my request by Dr. Morton's Clinic.

At one o'clock Saturday morning, September 10, 1994, baby Ammon made his appearance. He was our sixth child. The first few days it seemed to me that he was a little more restless than our other babies had been, but people said, "You just forgot how new babies can be."

At two days of age we took him to the doctor to be circumcised. The doctor examined him and mentioned he was slightly yellow. He told us to watch his color. At about four or five days old, he started clamping his mouth instead of feeding. He cried shrilly while arching his

back. Amazingly he slept better at times, which made people think he wasn't sick. As happened often with our other children in years gone by, we wondered, "Should we take him to the doctor?"

At ten days we decided something had to be done. I called our family doctor. There were no appointments available until two days later, so we would have to go to the emergency room to do something that day. I then called the local hospital. When they heard our situation, they told us to go to our family doctor and, "They will work you in."

After a long wait in the waiting room, the doctor finally examined baby Ammon. He didn't respond when the doctor slammed both hands on the cushion on which Ammon was lying. After consulting another doctor, he sent us to Russell Springs Hospital for a host of tests. By this time it was our usual bedtime. We decided I would go home and my wife, Miriam, would stay with Ammon. Miriam's sister was staying with our other children.

Ammon's temperature, blood count and heart rate were all low. An IV was hooked-up and heat and oxygen were turned on. Next a CAT scan was done, although it was not very clear.

By noon of the next day, Sept. 21, Ammon was transferred via ambulance to the University of Kentucky in Lexington, some 80 miles away. We followed several hours later by taxi. There they did more tests, and we needed a lot of patience and did a lot of waiting. Some tests were sent all the way to Texas. The results would not be available until after the weekend, and this was only Thursday. Family members and friends visited daily.

Thursday and Saturday night we slept at a Ronald McDonald House, which is provided for people who have a family member in the hospital. Things seemed kind of at a standstill, so Miriam went home on Friday night. I stayed alone that night.

On Friday night Ammon was moved out of the ICU, but by Saturday morning he was moved back because of vomiting and choking. At this point Ammon wasn't fully conscious and, of course, he was on medication, thus he didn't know whether I was there or not. I had wandered away for a few hours, and when I came back, Ammon was on a ventilator. The doctors said his breathing had dropped to six breaths a minute (approximately 1/10 of normal). How I wished Miriam would be here with Ammon, since he was gradually losing out. I now called the neighbors to let the family know the baby's condition and waited for Miriam and some others to arrive. Those faces certainly were a welcome sight when they finally came.

On Sunday, the baby still seemed to be losing out, but now I had Miriam and also my sister for support. Ammon's blood pressure dropped and some seizure activity was suspected. On Monday, Sept. 26, test results revealed maple syrup urine disease. As this was only the third known case in Kentucky, the doctor was unfamiliar with it. He called different doctors, who he thought might know about MSUD, but he didn't learn much. We finally got him to call Dr. Morton in Pennsylvania.

Dr. Morton asked our doctor if any of the other doctors had told him what to do. Our doctor said, "Not really." So Dr. Morton made some suggestions, but the University of Kentucky Hospital didn't have the special formula Ammon needed. So we contacted Leroy

Zimmermans from the western part of the state. They have a boy who has MSUD. Arrangements were made to have formula delivered yet that night by a local driver. According to the driver, they made the trip in record time.

It seemed to me it took the medical team a long time to figure out just how to mix the stuff. Finally, early Tuesday morning, Ammon was started on the formula. The lack of equipment to test the levels daily was the next obstacle which seriously slowed the treatment. Cincinnati, Ohio was the nearest place at which they could check the levels.

Now Dr. Morton recommended baby Ammon be moved again, this time by plane to the Lancaster General Hospital in Pennsylvania where Dr. Morton had facilities to care for him. Dr. Morton specializes in genetic disorders. Ammon was transferred by plane from Lexington, KY to Lancaster, PA Thursday evening, Sept. 29, the ninth day of doctoring. It took approximately three hours. On the same night Ammon arrived in Lancaster, Dr. Morton had the formula adjusted and Ammon off the ventilator. *(On arrival Ammon's leucine level was 22.3 mg/%, down from a high of 32 mg/%.)*

On Friday we traveled by van to PA to my sister Lena's in Bowmansville. That night Ammon vomited and drew some fluid into his windpipe, so once again he was put on the ventilator. By Sunday, he came off the ventilator for the last time. At this time his levels were normal. *(On arrival at the Lancaster hospital, Ammon was given Ketonix 2, MSUD Powder, and isoleucine and valine through an NG tube. More fats and calories were administered through an IV. In 48 hours his leucine blood levels had dropped to 2.8 mg/%.)*

Then Ammon got a yeast infection from his IV catheter, causing his temperature to go up. His IV was changed to his other hand and treatment begun. The IV was removed Oct. 7, and he was free of IVs for about a day. He still tested positive for yeast infection, so an IV was placed in his left leg to administer antibiotics. A week later the cultures still tested positive for infection. The most effective antibiotic was administered by IV.

By Oct. 10 Ammon was taking all his formula by mouth - he had been on a feeding tube. I neglected my diary during the next ten days. *(Ammon's leucine levels went no higher than 4.9 mg/% during the last two weeks of his hospitalization.)*

On Oct. 20, Ammon left the hospital. The month of hospital life seemed long, but the Lord surely helped us in many ways. We especially appreciated the visitors and phone calls. We had been staying nights with my sister Lena and had to make arrangements each day for transportation. One older couple took us to their home for the night. That was a welcome change.

After arriving back home in Kentucky, we had to learn how to care for Ammon - 700 miles from our doctor. All went reasonably well, although during Thanksgiving week we ran out of Diet Powder. We had waited until the last minute to order, and then the holiday interfered. The night before Thanksgiving Mead Johnson sent a man from Evansville, IN to deliver the Diet Powder. He had problems finding us, but drove in with the Diet Powder the next morning before we left for church. We had given up on him and arranged for travelers to bring some along from Leroy Zimmermans.

When Ammon was a little over a year old, he had a two-day hospital stay in Russell Springs. Pneumonia was suspected, but when he improved so quickly, they decided it must have been his bronchial tubes.

Ammon was the first MSUD baby in the Casey County settlement of Old Order Mennonites. The settlement was in existence about 18 years when Ammon was born. Two months after his birth, another child with MSUD, Curvin Zimmerman, son of Ivan Ray Zimmermans, was born. Three years later in 1997, Harold Zimmerman, son of Dana Zimmermans, was born and diagnosed with MSUD. Harold and Curvin are cousins.
Ammon, now a little over three years old, is doing fairly well. We generally have to keep pushing him to take enough formula and food. He does a little toe-walking due to the delayed treatment when an infant. He started to walk at about two years. His little brother Luke, our seventh child, joined us about that time. Thankfully he did not have MSUD.

UPDATE:
This history was sent to me at the end of last year. In January 1998, when in Pennsylvania, we heard that Ammon was in the Lancaster General Hospital. His parents were with him and we visited them there. Ammon was refusing to take anything by mouth, which was delaying his release from the hospital. The Shirks said they would bring the story they wrote earlier up-to-date. Following is Miriam's account sent to me in March.

On Christmas Day, 1997, we were at my sister's for a family dinner. Ammon didn't want to eat and wanted to be held all the time. He was running a fever. In the morning he had seemed OK. He still drank a little formula. Friday evening he didn't want to drink any formula. So John called the Clinic in Pennsylvania and talked with the nurse.

On Saturday, Dec. 27, John and I took Ammon to the emergency room in Russell Springs Hospital. Dr. Miles (our family doctor) ran tests to see what was causing his fever. His lungs were clear and the doctor couldn't find what was troubling Ammon. At four in the afternoon they decided to put in an NG tube. Ammon was admitted to the hospital for the night. I went home and John stayed with Ammon, who had a restless night.

On Sunday, December 28, Ammon was not improving, so Dr. Morton wanted him in Pennsylvania. We left home at 5 p.m.. Sunday and arrived at the Lancaster General Hospital Monday morning at 4 a.m. An MRI showed quite a bit of fluid on the brain *(brain edema)*. An x-ray showed the chest was clear. Dr. Morton still did not know what was causing Ammon's fever.

On Thursday, January 1, Ammon seemed happy and was singing "Precious Memories," but he wasn't drinking his formula. His DNPH was clear. His gums looked puffy and red, as if he had a sore mouth. He had a low sodium level and so they gave him a lot of salt. On the 4th, Sunday, they took out the IVs because they went bad. They put in an NG tube, and in that way he kept his formula down.

On January 9, Ammon was released from the hospital, and we stayed with friends in the area. He still had an NG tube, because we couldn't get him to take his formula. We had an appointment at the Clinic to see Dr. Morton on the 12th. On the 13th , with a lot of coaxing, we

finally got Ammon to drink some formula from his bottle. Using a syringe also, we were getting about half his formula into him.

We started for home on evening of the 14th and arrived home the next morning. We were still depending on the NG tube to get all the formula he needed into him each day. Now (March 1998), he is doing real well. He is hungry and eating at the table. He begs for his bottle which is something he rarely did before.

NEWS & NOTES - REPORT ON METABOLIC CONFERENCE

Details

Written by Sandy Bulcher

Published: 20 July 2009

Sandy Bulcher reports on the 1998 Metabolic Parent/ Professional Conference she attended in Massachusetts.

March 28,1998 was a beautiful Saturday for the New England Connection for PKU and Allied Disorders Conference. The meeting was held at the Holiday Inn in Taunton, Mass. Dr. Harvey Levy from Massachusetts spoke about newborn screening concerns. Some states are decreasing the number of diseases screened for. For this reason, he believes that we are "regressing rather than progressing" in the area of newborn screening.

Dr. Don Chase from NeoGen in Pittsburgh, Pa. followed with an explanation of mass tandem spectrometry. NeoGen has developed the technology to screen 30 diseases from one blood spot in less than two minutes. The company has screened 500,000 babies so far for both amino acid and fatty acid disorders. John Sarrantino, former Executive Director of the New England Screening Program, spoke about the need to educate government officials about newborn screening issues.

Dr. Holmes Morton from Lancaster County, Pa., discussed the benefits of early diagnosis, especially for MSUD and Glutaric Aciduria (a metabolic disorder common among the Amish population of Lancaster County). He used powerful visual aids to illustrate the difference between children who had the benefit of early diagnosis and those who did not.

Dr. Mark Batshaw from Pennsylvania spoke about gene therapy. Although researchers have identified the virus to use with the gene, problems still exist in getting the gene into the body and keeping it working. Dr. Batshaw has done work specifically with OTC Deficiency (a urea cycle disorder) and hopes in five years to use gene therapy in children with OTC Deficiency.

Prior to lunch, we divided into groups by specific disorder. Families representing PKU, MSUD, Homocystinuria, Organic Acidemias, and Urea cycle disorders were all present. I was excited about the number of MSUD families present. Richard and Sheila Pollica from Vermont, John and Rachel Ennis from Massachusetts, Paul and Cheryl Needleman from Massachusetts, Ana Montalban from Rhode Island, and I met informally with Dr. Morton to discuss issues pertaining to MSUD.

Following lunch, Dr. Levy updated the PKU families on PAL, an enzyme that was developed with the theory that it would allow PKU children to ingest increased amounts of phenylalanine. The research is on hold, however, because phenylalanine levels did not decrease as much as predicted.

The coordinator of the conference, Trish Mullaley, updated everyone on the Scott C. Foster Metabolic Research Fund. The organization has set a goal of $500,000 for research for metabolic disorders and has already raised $120,000.

During the afternoon break, families had time to purchase products, talk with vendors, sample low protein products, and meet new friends. Finally, a panel discussion with Dr. Levy, Dr. Morton, and Dr. Batshaw completed the afternoon. Dr. Morton expressed concern that fewer physicians are being trained in the field of biochemical genetics, thus fewer will have the expertise to care for children with metabolic disorders. Dr. Levy informed us that the best means of educating physicians and lay people about metabolic disorders is through newborn screening programs. Undoubtedly, everyone who attended recognized the importance of improving newborn screening programs nationally, and felt empowered to make a difference.

NEWS & NOTES - NATIONAL PATIENT AIR TRANSPORT HELPLINE

Details

Written by Joyce Brubacher

Published: 20 July 2009

Mercy Medical Airlift (MMA) sent an article to reprint in newsletters describing their services. Since I wasn't sure how it could be utilized by our families, I called the helpline number and received the following information. This service is not ordinarily available for emergency situations because the volunteer pilots are working persons and need advance notice. However, this could be a valuable service for those families who have limited funding and need to travel several hours or more for their clinic appointments.

The National Patient Air Transport Helpline (N-P-A-T-H)

1-800-296-1217 is a one-of-a-kind unique hotline which makes referrals to all known appropriate charitable, charitably-assisted and special patient discount commercial long-distance air medical transport options. By calling the 24-hour helpline you can tap into a wealth of available resources. Referrals are based on an evaluation of the patient's medical condition, type of transport required and departure/destination locations. Patient referrals are made to over 45 different sources of air medical transport help.

N-P-A-T-H provides information and referral for:

- Those who must find a way to travel considerable distances for specialized evaluation, diagnosis, treatment, rehabilitation, and/or recovery necessitated by illness or accident.
- Q Health care professionals (doctors, nurses, social workers, and discharge planners) who must find the most cost- effective way to move patients and their family members long distances.
- Q Volunteer pilots who want to serve with one of the volunteer pilot agencies that is part of the Air Care Alliance (ACA). ACA is the nationwide league of humanitarian volunteer pilot organizations flying for public benefit.
- Q The "special lift" air medical transport program operated in conjunction with N-P-A-T-H. Sponsors of large-scale disease research or experimental treatment programs can take advantage of this program which will manage and coordinate the air medical transportation aspect of the special project - arranging to move large numbers of patients to and from special research or treatment facilities, one at a time, as required (via charitable means) nationwide.

NEWS & NOTES - THE SPECIAL KID'S NETWORK (FOR PENNSYLVANIA FAMILIES ONLY)

Details

Written by Joyce Brubacher

Published: 20 July 2009

The Department of Health's Special Kids Network (SKN) is a statewide information service assisting parents, agencies, or anyone in Pennsylvania seeking help with a child who has a chronic or disabling health condition. By calling 1-800-986-4550 from anywhere within Pennsylvania, a caller is automatically routed to the closest of six regional offices throughout the state. Trained telephone counselors respond with information and referrals obtained from a comprehensive, statewide database. The database within each region includes agencies from that region as well as statewide and national agencies providing

over 103 distinct services grouped into six broad categories: education/training, healthcare products/services, recreation/ leisure, social service/counseling, support/advocacy, and therapy. There are over 5300 agencies in the SKN database.

The goal of the SKN is to make connections between families of children with special health care needs and providers offering services. The SKN specifically targets children under the age of 21. SKN staff members are available to provide information or make presentations about the Special Kids Network and its related services. To arrange for a presentation or request additional information, please call 1-800-986-4550.