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Spring/Summer, 1999

NEWBORN SCREENING COALITION

Details

Written by Joyce Brubacher

Published: 20 July 2009

The New England Connection for PKU and Allied Disorders held their 1999 PKU Parent/ Professional Conference in Tauton, Massachusetts on March 20. Sandy Bulcher represented our MSUD Family Support Group at the conference. On the evening of the 19th, a group of persons interested in newborn screening met to discuss issues relating to state screening programs. Here Sandy reports on this meeting. (More information on the meeting on the 20th farther on in this issue.)

I've been interested in the issue of newborn screening since our son Jordan was diagnosed with MSUD nine years ago. At that time, it was painful to learn that had we lived in one of the 21 states that screen for MSUD, Jordan would not have had to suffer 17 days before a right diagnosis was made. Over the last few years, I have been trying to have MSUD added to Ohio's newborn screening program, but had never expanded my thinking beyond that. Fortunately, Trish Mullaley, a PKU parent and former president of the New England Connection for PKU and Allied Disorders, had a vision to improve newborn screening on a national level.

Last fall, we started making plans for the first gathering of parents and professionals from across the country interested in newborn screening. We were able to see this vision become a reality on Friday, March 19, 1999 in Taunton, Mass. Approximately 25 parents and professionals met to discuss flaws in our current newborn screening system, as well as the need for expanded screening nationally. Parent representatives from the following organizations attended.

- PKU of Florida
- PKU of Illinois
- Children's PKU Network (California)
- Mid Atlantic Connection for PKU
- PKU Organization of Wisconsin
- Ohio Coalition for PKU and Allied Disorders
- New England Connection for PKU and Allied Disorders

- MSUD Family Support Group
- Parents of Galactosemia
- Homocystinuria (family representative)
- Organic Acidemia Association

In addition to these parent representatives, professionals from several formula companies, a low protein food company, and two newborn screening companies were present. We were honored that Dr. Richard Koch from California took interest in our group and attended.

I've learned a lot since researching the issue of newborn screening. First of all, even though all 50 states and the District of Columbia screen for PKU and Congenital Hypothyroidism, this does not necessarily detect all these infants, and numerous infants are missed annually. Frequently, the problem occurs because of the lack of education, poor communication, or inadequate follow-up. We discussed numerous ways to address these problems including printing educational materials, developing a Web site, and seeking the support of organizations such as the National Institutes of Health and Center for Disease Control.

Except for PKU and Congenital Hypothyroidism, states vary in the diseases for which they screen. *(Only twenty-one states screen for MSUD at this time.)* In the last several years, new technology has been developed, *tandem mass spectrometry* (MS-MS), which has the capability to test for approximately 30 diseases including MSUD. Several states are using this new technology for newborn screening in addition to their current program. All present at the coalition meeting felt that promoting expanded screening through MS-MS would be beneficial, in addition to addressing problems in our current system.

Also, everyone agreed to forming a national non-profit organization called the National Coalition for PKU and Allied Disorders. Initially, we will be looking at newborn screening issues, but will expand into issues related to insurance coverage for formula and low protein foods. The coalition will be set up so that a representative from each of the metabolic organizations can obtain information and disseminate it to their local organizations. I'm really excited about being involved with the National Coalition and feel confident that we can make a difference. I will keep you informed of our progress. Thanks to SHS, Wallac, and Neo Gen for funding this first important meeting.

Sandy is involved in legislative efforts to improve newborn screening in Ohio and on the national level. Contact her for information: Sandy Bulcher, 9517 Big Bear Avenue, Powell, Ohio 43065. Phone: 614-389-2739 E-mail: dbulcher@juno.com

NEO GEN SCREENING

Details

Written by Joyce Brubacher

Published: 20 July 2009

Neo Gen Screening, Inc. is a laboratory specializing in neonatal screening and the diagnosis of inborn errors of metabolism. The lab has analyzed over a half million samples for inherited metabolic disorders using tandem mass spectrometry and other biochemical genetic methods. Other specialty and diagnostic testing services are offered including urine organic acid testing and quantitative amino acid analysis. More than 30 clinically significant and manageable diseases that affect newborns are tested for in their Supplemental and Comprehensive Newborn Screening Program.

Automation and high volume testing in their routine newborn screening laboratory allow them to offer programs and specialty testing services at considerably lower prices than currently available in other laboratories. They will provide additional information on request detailing their clinical and newborn screening programs and the procedures for specimen collection, shipping and handling, pricing and billing information.

Free screening packages are available for clinical metabolic screening services. The package includes a Neo Gen Screening filter paper for collection of specimen, a glassine envelope, a pre-paid pre-addressed envelope, a list of disorders, instructions for specimen collection and order forms for additional "free" testing packages. Screening is available for 30 diseases at a cost of $25.

Contact: Neo Gen Screening, 110 Roessler Road, Pittsburgh, PA 15220-1014. Phone: 866-4636436 Fax: 412-220-0784 E-mail: info@neogenscreening.com Web site: www.neogenscreening.com

NEWBORN SCREENING IN NEW ENGLAND

Details

Written by Harvey Levy, MD

Published: 20 July 2009

Reprinted from the New England Connection Update, March 1999, a newsletter of the New England Connection for PKU and Allied Disorders. This article provides a brief history of newborn screening and explains the importance of tandem mass spectrometry.

New England has traditionally been the center of action in newborn screening. In fact, newborn screening really began in New England. Dr. Robert Guthrie (Buffalo, New York)

had developed a newborn screening test for PKU, and he knew that it could prevent mental retardation in thousands of children with PKU. However, the medical and public health establishment in New York at the time opposed making the test routine for all newborn infants.

Fortunately at a meeting in 1962, Dr. Guthrie met Dr. Robert MacCready, Director of the Diagnostic Division of the Massachusetts Public Health Laboratories. Dr. MacCready learned about the screening test from Dr. Guthrie, was convinced that it would indeed prevent mental retardation in PKU, brought the test to Massachusetts, and asked doctors throughout the state to obtain heel stick blood specimens from newborn babies. This became the first newborn screening program in the world. In 1963 Massachusetts became the first state to pass a law requiring a PKU test in all newborn infants. Other New England states soon followed Massachusetts, and this was followed by still other states throughout the country.

When Dr. Guthrie developed other tests for disorders such as HCU, MSUD and galactosemia, he looked to Massachusetts and other New England states to add these to newborn screening. When I became involved in the Massachusetts newborn screening program, New England had already become world famous as the center of newborn screening activity. This fame continued until a few years ago when other programs began to use new methods that expanded the number of disorders that could be identified and made the screening much more efficient.

By far the most important of these methods is tandem mass spectrometry, often abbreviated MS-MS. This method was pioneered by Dr. Don Chace when he was at Duke University and applied to routine newborn screening by Dr. Edwin Naylor in Pittsburgh. Dr. Naylor has now formed a private newborn screening laboratory known as Neo Gen Screening, Inc., where he and Dr. Chace are now screening almost 200,000 babies a year by this method. They have shown they can identify many infants with important disorders in addition to PKU and prevent disability or even death in these infants by beginning early treatment made possible because of newborn screening.

Getting MS-MS adopted in New England, however, proved to be a formidable task. By this time the individual New England states had decided to have a regional newborn screening program with the screening performed in only one laboratory, the one in Massachusetts. All New England states except Connecticut are part of this program; Connecticut preferring to retain its own testing laboratory. Thus, advocacy for adding MS-MS to newborn screening had to center on Massachusetts. The metabolic doctors in Massachusetts, including Dr. Mary Ampola, Dr. Mark Korson, Dr. Vivian Shih and myself, vigorously fought to have MS-MS adopted here.

The matter was hotly debated within the Newborn Screening Advisory Committee of the Massachusetts Department of Public Health. Fortunately with allies such as Dr. Philip Reilly, Director of the Eunice Kennedy Schriver Center, and with the very strong support of the Connection led by Trish Mullaley, we were able to get the Public Health Department to pass new regulations that established MS-MS screening on a 2-year pilot basis. This screening program began in Massachusetts on February 1, 1999. We are confident that this expanded screening will continue once its many advantages are realized. We are also confident that

the other New England states will soon add MS-MS. If so, New England will once again become a world leader in newborn screening.

I cannot emphasize enough how important parents and parent organizations are in this process. As I have mentioned, the Connection has been critical in helping the Massachusetts laboratory turn in the right direction. ***Now needed is an effort to make newborn screening consistent throughout the U.S.***, so no longer will identifying a disorder in a baby depend on which state the baby is born in. The federal government is finally beginning to take a role in this matter, but constant vigilance both within individual states and at the federal level will be required to assure appropriate final results.

ROBERT GUTHRIE: THE PKU STORY

Details

Written by Joyce Brubacher

Published: 20 July 2009

Review by Joyce Brubacher

Wayne and I attended the 1999 PKU Parent/Professional Conference in Massachusetts in March. We met Dr. Richard Koch from Children's Hospital in Los Angeles and his wife, Jean. Dr. Koch spoke on "Positive Results of Late Treated PKU" at the conference. He has been very involved in treating PKU for many years. He and Jean were very good friends of Dr. Robert Guthrie. In the early 60s, Dr. Guthrie developed the bacterial inhibition assay used for screening for PKU and MSUD. If it had not been for Dr. Guthrie, persons with MSUD would have a much greater risk of damage and death today.

Jean Koch wrote a biography of Dr. Guthrie, *Robert Guthrie: The PKU Story*. She tells the story of Bob Guthrie as a family man as well as a scientist. The father of a retarded son, Dr. Guthrie dedicated his life to the prevention of mental retardation. His crusading spirit led him to present programs throughout the world on the need for the early detection of conditions leading to mental retardation. In 1968 he took his wife and children on an adventure around the world, camping out with their two VW buses in various countries, sightseeing and visiting medical centers to encourage newborn screening.

Jean was kind enough to autograph my copy of her book. I have found the biography quite interesting. Our own interest in newborn screening also started in the 60s when our son Monte was born in Oregon. He was the first infant in the world with MSUD to be picked up through a state screening program. Dr. Guthrie flew to the state of Oregon when Monte's second Guthrie test was confirmed positive. Monte was making medical history, and Dr. Guthrie was the reason it was possible.

It was many years later that we met Dr. Guthrie personally. Then in 1988, he and his wife Margaret attended our Symposium at Hinkeltown, PA. They stayed for the full symposium. He took an interest in our support group and was very encouraging.

Dr. Guthrie's dedication to preventing mental retardation was instrumental in saving the life and health of our son in 1965 and many others with MSUD since. This book reveals the *real* Dr. Guthrie and the many obstacles he overcame to leave this contribution to mankind. I highly recommend it to those interested in the history of newborn screening, medical history or general interest in a dedicated father and scientist.

The book is available from your local bookstore or can be ordered from:
Hope Publishing House
P.O. Box 60008
Pasadena, CA 91116
E-mail: hopepub@loop.com
Web site: www.hope-pub.com

GLEANINGS FROM THE NEW ENGLAND CONNECTION CONFERENCE

Details

Written by Joyce Brubacher

Published: 20 July 2009

Editor's Observations
The New England Connection for PKU and Allied Disorders held their 7th annual PKU & Metabolic Conference in Massachusetts for parents and professionals on March 20, 1999. Wayne and I attended and appreciated the presentations.

There are many similarities in PKU (phenylketonuria) and MSUD. One of the biggest differences in the past has been in dietary control. In the 60s and 70s, most clinics allowed those with PKU to go off diet when they were around six years old. Only a few clinics thought it necessary to maintain the diet for life.

Today it is evident that those with PKU, who stop the diet, gradually suffer the toxic effects of elevated levels of the amino acid, phenylalanine. These problems have only recently been recognized to be related to elevated levels in those who were early treated. Reports indicate lowered IQs, mood alterations, impulse control disorders, migraine headaches, lack of ability to

concentrate, loss of short term memory, poor academic functioning, inability to hold a job, muscle weakness, tremor and poor social and personal relationships.

These problems vary in individuals with PKU. However, those who go back on diet or maintain the diet from infancy, find these conditions disappearing or greatly improved. There are adult persons with PKU with high IQs and in professional positions. Scott Merrill, an attorney at law, has PKU and was a master of ceremonies at this PKU meeting. He went back on diet as an adult and found his academic abilities improved.

Can we apply this observation to MSUD? I am hearing from adult persons with MSUD who are having some of these same problems. Although these adults were on diet since infancy, in the 70s and 80s it was not common practice to maintain leucine levels as low as recent evidence indicates best. Today improved treatment, including the addition of isoleucine and valine to daily diets, appears to help maintain lower and more controlled levels even in adults with MSUD.

The make up of individuals certainly plays an important part in the way high levels affect them. It is difficult to know for certain the cause and effect relationship of physical and mental problems to MSUD. Which effects are related to the time treatment began or to lack of dietary control or other causes? There are reports of headaches, sensitivities, body aches, tremors, social problems, inability to reason adequately, excessive talking, poor academic functioning, fatigue and weakness in muscles and joints. Some adults with MSUD may not experience any of these problems. As with PKU, some individuals may not recognize that their problem is similar to others with the same disorder.

At the PKU meeting there was a strong emphasis on maintaining diet for life. I would like to also encourage very strict dietary control for all those with MSUD regardless of age. Don't compromise future mental and physical health.

I am very interested in more documentation on the effects of MSUD on adults. I encourage those with MSUD and their parents to contact me. All information will be kept confidential.

Engineered Protein

Imagine adults with PKU going back on diet and facing the challenge of drinking the less-than-desirable tasting (to put it mildly) medical formula for the major part of their diet. As with MSUD, those on the PKU diet keep wishing for more normal foods. The many new low protein products help but they cannot substitute for the medical formula which contains needed protein.

One person responding to this plea for a more normal diet is Dr. Bryan Hainline from the Department of Pediatric Metabolism and Genetics, Indiana Medical Center, Indianapolis, Indiana. He spoke on "Development of a New High Protein Phe-Free Diet Supplement" at the PKU Conference. (Phe is the abbreviation for phenylalanine, the amino acid involved in PKU and one of the essential amino acids found in all protein.) Following is a summary taken from this speech and an article on the same topic by Dr. Hainline in the *PKU News* (Vol. 10, No. 3, Winter '99 issue).

Dr. Hainline and his associates became interested in developing a phe-free protein which can be added to low protein foods. This would add a high quality protein to the diet in the form of foods that can be baked and heated and have the protein content of "normal" foods. Commercial companies could make high protein-low phe chips, breads, pastas, cookies, etc. By eating foods containing this new protein, persons with PKU could reduce the amount of formula they need because they would be getting some of their phe-free protein from these foods.

Using medical genetic techniques, they proposed to remove the phenylalanine from a corn protein called gamma zein found in the kernel of the corn. This protein contains 203 amino acids, two of which are phenylalanine. They have already modified the gene that codes for the production of gamma zein, allowing them to remove the two phenylalanine residues. Now they need to improve the nutritional content of the protein and produce the phe-free protein in a plant.

This naturally occurring gamma zein protein purified from corn does not have a distinctive flavor so is expected to take on the flavor of any food it is mixed with. Gamma zein is related to gluten, the protein in wheat that gives food baked with flour much of its structure. Therefore, it has an excellent chance of improving the texture of low protein foods. To be effective, large quantities of this protein would need to be eaten, so it is expected to be more useful for older persons with PKU. Other nutrients found in the formula would have to be supplied with vitamin/mineral supplements and additions of dietary fat and carbohydrates. The new foods could only be used under medical supervision.

If this research is successful in producing a phe-free protein, the underlying technology will be useful for designing similar proteins for other inborn errors of metabolism including MSUD. The projected time for this new protein to become available is five years according to Dr. Hainline. However, he recently told me that a lack of funding is hindering the research. He is currently looking for personnel to work in the laboratory. We can only hope the research in gene repair therapy discussed in the following article is successful so we may not need genetically changed protein for MSUD.

NOTE:
The Scott Foster Metabolic Research Fund has supported Dr. Hainline's research. It is the type of research for which the fund was established - research which can benefit a number of metabolic diseases including PKU and MSUD. However, much larger sums of money are needed to make an impact. To donate to The Scott Foster Metabolic Research Fund, send your donation to:
The Scott Foster Metabolic Research Fund
New England Connection for PKU and Allied Disorders
16 Angelina Lane
Mansfield, MA 02048
Phone: 508-261-1291

Update on Gene Repair Therapy
Dr. Blaese spoke on "Are We Getting Closer to Gene Therapy?" at the PKU Conference. (Michael Blaese, MD is Chief of Clinical Gene Therapy Branch, National Institute of Health and President of Kimeragen Molecular Pharmaceuticals.) In the Fall/Winter '98 *MSUD*

Newsletter, we reported on the technology of gene repair therapy as explained by Dr. Blaese at Symposium '98.

Dr. Blaese explained the difference between this new gene repair therapy and the traditional gene therapy. Gene repair actually corrects the gene code by using a chimeric molecule (the chimeraplast) to harness the cell's normal DNA repair system to correct the gene.

Certain diseases cannot be corrected, only those with single cell mutations. Dr. Blaese gave a list of ten diseases that are targeted for this gene repair. MSUD and PKU are both on this list. However, the first disease scheduled for human trials is Crigler-Najjar - a very rare mutation in an enxyme required to properly metabolize and eliminate bilirubin. The bilirubin levels cause organ damage and the child often dies in the second year of life.

I asked Dr. Blaese for an update on the first human trials and his response follows.
"Progress is moving well toward our goal of beginning treatment for the Crigler-Najjar children before the end of this year. The clinical protocol is now being written and the documents needed for FDA review are being assembled. Last week we made our final determination of the exact structure and formulation for the chimeraplast that we will use for the Crigler trial, and we have now started to manufacture the larger amount of pharmaceutical grade material needed for the trial. We expect to submit our formal application to the FDA after all the required safety testing is complete in September and hope to begin treating three patients by November. However, we still have a lot of work to do before I am comfortable enough to actually begin treating the children. We do not want to begin prematurely or potentially cause harm, so it is possible that this preliminary schedule could be bumped back if issues come up that take more time to resolve.

Kimeragen was able to find some investors who provided funding for several months, and we are hopeful that additional money will become available so we can complete the initial trial - and demonstrate to the world that this unique therapy really does have the potential to change people's lives for the better. We are also looking for ways to obtain grants and contracts to support development of treatments for other orphan diseases, such as MSUD, and we are absolutely committed to finding a way to bring this treatment to those other conditions."

NEWS & NOTES - BOOKS REVIEWED

Details

Written by Denise Pinskey

Published: 20 July 2009

I received a request through Joyce Brubacher concerning books that may help MSUD families cope with MSUD issues. The request was made by Lori Smith Thornton, Aunt of Kayleigh Hahn. I called Magination Press so I could review any books I felt would help our situations. Two books sounded helpful.

What about Me? When Brothers and Sisters Get Sick
by Allan Peterkin, M.D.
In this book you are introduced to a family with a daughter and a younger son. It is written from the sister's perspective. It tells how the sister really likes having a little brother. But when her brother does something to upset her, she wishes she never had a little brother. Unfortunately her brother becomes tired and cranky, and has to go to the hospital. The story does end happily with the parents talking with the doctor and the sister. And the brother does get better.

I found this book to be very helpful with my four-year-old daughter who sometimes feels very left out when Zachary (our son with MSUD) gets sick. We had read this book once while Zachary was in therapy. A few days after reading this book, Zachary became ill and our four-year-old referred back to the book in explaining why Mommy and Daddy had to take Zachary to the hospital, and she had to stay at Grandma's house. So for us, this book was helpful in starting conversations with her.

Little Tree: A Story for Children with Serious Medical Problems
by Joyce C. Mills, Ph.D.
In this book you are introduced to a happy tree, Little Tree and a squirrel, Amanda. During a storm, Little Tree's branches are hurt and Amanda calls upon the Tree Wizards of the forest to help. Unfortunately, they can't fix Little Tree's branches. They have to remove them in order to save her life. With Amanda's help, Little Tree learns to accept her new, changed self and to find other strengths inside herself. The story also introduces the practice of the "Magic Happy Breath." This is where Little Tree had to take a deep slow breath and imagine the beautiful sky swirling through its whole trunk and branches. I think this practice would be helpful to relax your children when they get upset over medical procedures.

If you are interested in either of these books or want a list of all the books available from Magination Press, you can visit their Web site: www.apa.org/books, or call 800-374-2721 or write to Magination Press, American Psychological Association, P.O. Box 92984, Washington, DC 20090-2984.

NEWS & NOTES - SCHOOL HONOR

Details

Written by Joyce Brubacher

Published: 20 July 2009

Melissa Berman, 17, daughter of Bob Berman, was named Student of the Month for the Physical Education/Health Department at Peabody Veterans Memorial High School, Peabody, Massachusetts. She received a certificate of recognition, a Peabody Tanner Pride Pin, a bumper sticker for her dad, gift certificates for combo meals at a new food concession, and a free buffet luncheon with her nominating teacher. Additionally, her picture

and a narrative explaining why she received this special honor appeared in the Student of the Month display case at the school. She was applauded for her hard work and commitment to her studies. This was quite an honor in a high school with 2000 students. CONGRATULATIONS FOR A JOB WELL DONE, MELISSA!

NEWS & NOTES - FAMILIES MEET IN OHIO

Details

Written by Joyce Brubacher

Published: 20 July 2009

Twenty MSUD families in the Michigan, Ohio, Indiana, and surrounding area were interested in getting together. It was decided to have a fun weekend of sharing at a central location. Denise Pinskey reserved rooms for families at a Howard Johnson motel in Perrysburg, Ohio. The price was good and the pool was just what the children needed on a this hot day, April 17, 1999. The seven families who were able to attend really enjoyed that weekend.

We would encourage families in other areas to get together and share. It takes some effort to plan, but is well worth it. Maybe your area has only a few scattered families. The number isn't important, just get together. It may be two or more families visiting the zoo or just an afternoon at the park. It means so much to the children to share with others who have the same diet which could include families with PKU children.

CARING IS SHARING.

NEWS & NOTES - LATE DIAGNOSIS, WHY?

Details

Written by Joyce Brubacher

Published: 20 July 2009

Since we have a Web site, we hear about persons with MSUD from all over the world and have been able to help them in various ways. Several recent countries we have heard from are Brazil, South Korea, New Zealand and Israel. It is so sad that children with MSUD die undiagnosed or are critically ill before being diagnosed in many countries.

What rationale is there for this still happening in the United States? Every state has a newborn screening program for at least PKU. At very little additional cost, all newborns could be screened for MSUD as well as a number of other diseases. In May of this year, a child was in the hospital in Florida for two weeks in critical condition until diagnosed at four weeks of age with classic MSUD. This should not happen. This issue needs to be addressed. If you are interested in helping, contact Sandy Bulcher (see more information and how to contact Sandy under NEWBORN SCREENING COALITION at the beginning of this issue).

FORMULA SUGGESTION

Details

Written by Sanse Swenson Winstead

Published: 20 July 2009

I have been drinking my formula for 28 years, and over the years I have tried many different brands of formula and used many different ways to measure and mix. The latest method seems to work the best for me.

I now measure and mix my formula for each *serving* and not for each day. Have you ever seen those coffee cups at McDonalds? They are Styrofoam cups, with a plastic lid. The lid has a little tab that you pull back, making it into a sipping cup, blocking that smell. I purchase the same type of cups and lids from Sam's Warehouse by the case, and they are not very expensive. What I do is measure enough powder into a cup for a serving. I usually have four servings per day. When it is time for my "power drink," I grab a cup containing premeasured formula, add water and mix it with a little battery powered hand mixer, drink my formula, and toss the cup in the trash.

I don't have to wash blenders, mixers, containers, cups, coolers, etc. anymore. My formula is always fresh . . . made and drunk within minutes. I don't carry already mixed formula anymore, and it is very liberating. The little hand mixer can fit easily into my purse. The mixers can be purchased at Walgreens or a health food store for about $3. So I now can travel with formula without all of the baggage!

I drink the Complex Vanilla formula and I highly recommend it. It smells better, tastes better with no bitter aftertaste. Sometimes, no matter how easy it is to carry the formula, it is still an inconvenience. For those times, I use the Amino Acid Bar. Let me know what you think.

Shayla also takes the Complex MSUD medical food from Applied Nutrition that Sanse writes about. She likes it so well she says she could eat it dry out of the can. She did not like any of the previous formulas she tried. She also finds the Amino Acid Bars very handy when traveling or gone from home for a day. Maybe Sanse's idea will be a help to others struggling with formula problems.

The Complex Amino Acid Bar, a chocolate bar marketed by Applied Nutrition, contains 10 gm amino acids (no leucine, isoleucine or valine). It can be used to substitute for part of the formula, but the nutritional value is not the same. Consult your nutritionist before using.

Cans of medical food always say to refrigerate after opening. I asked the company about refrigeration, and it is not necessary to refrigerate in the can, only after mixing with liquid. However, it tastes best when mixed as a single serving and drunk immediately, just as Sanse does. It is important to protect the formula from humidity. Moisture in a can of dry formula can make the powder cake, and may turn it an ugly color. It does not, however, harm the potency of the formula.

SHARING - THE RUTER FAMILY'S "BIG APPLE" ADVENTURE

Details

Written by Paula Ruter

Published: 20 July 2009

Perhaps some of you were able to see the November 27, 1998 airing of the Sally Jessy Raphael show. My husband Lance, my daughter Anna, and I were guests on the show. We were there to discuss MSUD. I am writing to share our experience of being on a national talk show.

I will confess, I do occasionally watch talk shows. Sometimes one needs to know that there are people out there who have more turmoil in their lives than you. Anyway, back in August while exploring the Internet, I looked up the Web site for Sally's show. There was a spot to suggest show ideas. I suggested that a show should be done to explore rare diseases.

This was all forgotten until I received a phone call one Thursday in late October. It was Ashli, a producer for the Sally show, and she wanted to know more about MSUD. After I explained the disease and our experience with it, she told me they were planning on doing a show about rare medical disorders. She said she had to talk to Anthony, another producer, and that I could expect a call from him the next day.

I really did not want to get my hopes up that we would be asked to be on the show, but my heart jumped every time the phone rang. Finally, Anthony did call. He said they were taping the show on the following Wednesday, and they wanted us on it! I was amazed! Us, on a talk show! Lance and I were so excited!

We were also very nervous. At the time, Anna was fifteen months old. How would she handle a plane ride? We also had an added challenge. Anna had broken her leg in September and was in almost a full body cast. As luck would have it, Anna's a born traveler. They flew us out to New York City on November 3. Anna loved the flight! She either slept or flirted with the business travelers.

Once we made it to New York we were treated like celebrities. We were picked up at the airport in a beautiful limousine. We just stared, wide-eyed at all of the landmarks. The Empire State Building was just beautiful, lit up in the night sky. We were *really* wide-eyed when we pulled up to our hotel. We stayed at the Le Parker Meridien, a gorgeous four diamond hotel on 57th Street. I wonder what the people on the sidewalk thought when a limo pulled up, but we were the ones who got out. We stayed in a suite that had a nightly rate higher than our monthly rent at home.

Unfortunately, we did not have a chance to enjoy the city. We were picked up for the taping at 6:45 the next morning. Since we were scheduled to be the last segment on the show, we had a long wait. We shared a room with another family. Their baby suffered from a disorder where she is covered with giant moles. We also met Ellen Mae, a thirteen-year-old girl with Freeman Sheldon Syndrome, an orthopedic disorder. However, we never actually met Sally until we were taping.

Finally, it was time for our segment. We were fitted with microphones and shown to our seats on the set. I have had many people ask me if we were nervous. It wasn't until we were sitting there, looking at the audience and seeing Sally standing in front of us. Yet, as you have all experienced, explaining MSUD becomes a way of life. Once Sally started asking questions, I felt as comfortable as if I were talking to someone in the grocery store. Poor Lance was not able to get a word in edgewise, though. Sally directed her questions to me, and as soon as I was finished talking, she went to the next question. Anna did wonderfully during the taping. As much as she loved the applause and the cameras, she may be destined for a career in show business! The taping seemed like it was over before we knew it. Then we were whisked back into limos, and sent to the airport. Our big adventure in the "Big Apple" was over, and it seemed like it had just begun.

About four weeks later, the show finally aired. We were very disappointed, because a large portion of our segment was edited out, to make up for time. The major reason we decided to go on the show was to advocate newborn screening. In our original taping, I spoke about how lucky we are that Anna was born in one of the 21 states that screen for MSUD. We pressed the fact that early detection is a key factor in controlling this disease. Yet that segment was completely taken out, along with a few others.

Despite that, we do not regret doing the show. We have had people approach us at church and at the store where Lance works who want to know more. Of course, one of the first things they say is, "You wouldn't know by looking at her." We are glad to be able to educate people on what MSUD is.

This was also a once in a lifetime experience. Well, maybe twice - we would still like Rosie O'Donnell to call! Once Anna is older, this will be something that she can be proud of.

SHARING - A SPECIAL DAY FOR CHELSEA AND NOAH

Details

Written by Kimberly Patterson

Published: 20 July 2009

Kimberly Patterson is the mother of two children with PKU, Chelsea 8 and Noah 7. The children live an active and normal life, both involved in various sports and other activities. Kimberly always thought the toddler years would be the most difficult with the associated dietary problems. But she found it more difficult to have the children in school. She met the problem head-on by sharing information about PKU with her children's 1st and 2nd grade classmates. Her excellent ideas are very applicable to MSUD. Following is a condensation of her article from the March '99 newsletter of the Association for Neuro-Metabolic Disorders.

It was tough for me to see my children's heartbreak over being "different." At times my PKU children have expressed feeling powerless regarding their diet and lack of diet choices. They have voiced such things as:

- "Mommy, some kids at school say that my milk stinks."
- "I wish I could eat what the other kids at school have so that I am not different."
- "When I bring plain pickles or mushrooms or whatever to lunch, the kids say, 'Ooh, that's gross!'"
- "I wish I didn't have to bring my milk (Phenyl-Free) to school."
- "Today I felt left out because Justin brought a birthday treat to school and I couldn't have it. It had nuts in it."

I have heard it said, "Knowledge or understanding is power." So I decided to give more "information" to Chelsea and Noah's peers by way of a "PKU Presentation." I went to their school classes armed with resources on PKU and ready for questions. Their teachers were more than happy to let me come in and share with their classes.

This is what I did:

1. I read a story to the class titled *Denny the Dragon and his Magic Milk* by Nancy Beiman, Maria Rosetti, and Holly Wolf. This is a story about a dragon with PKU. As I read the story, I picked opportune points in it to relate to my children's life and diet. It is printed in black and white similar to a coloring book. I made copies of each illustration in the book and distributed them to their

classmates and requested that they each color a page and sign it, so that my children would have a memory of their class as they read their book.
2. I showed them a photograph of our family (we have 2 PKU and 2 non-PKU children) and talked about genetics a little - well, as much as you can with 1st and 2nd graders.
3. I took out the low-protein food list and explained a little more about counting Phe's. I let the children choose food items that they were curious about, and then showed them how to look them up and find their Phe value.
4. I showed them two of our low-protein cookbooks and explained that I have to prepare some of their foods with special ingredients that we order through the mail.
5. I brought a can of Phenyl-Free and passed it around the room, letting the children smell and feel it. We talked about how we prepare the formula each morning and how important it is for Chelsea and Noah to drink this every day if they want to feel good.
6. Then we opened the floor for questions. The children were so insightful and curious. What a delight to see my children answering their classmates' questions with confidence and pride!
7. We showed the class the diet record that we had been keeping for the previous two days. Then I explained how we send the records in with a blood spot to PKU clinic's lab in Ann Arbor.
8. I took Chelsea and Noah's blood sample right there in class, so that their friends could see what they go through. Both of my children were a bit nervous about this so I reassured them that they did not have to do it, that we could wait until later in private. But, they both decided to go ahead. Their friends were so great! Chelsea's girlfriends were very empathetic and gave her hugs afterwards. Some of them asked questions like: "Did it hurt lots?" and "Were you afraid?"

Both classrooms had such a great response to the presentation. The teachers and assistants had really great questions too. My children both rushed in the house that afternoon after they got off the bus with smiles on their faces and comments like: "Mommy, everyone thinks I'm famous now." "I think my friends in class understand PKU now." What a return for just a small investment of my time! I only wish I had done it sooner.

If any of our MSUD parents try this idea, or have used another other ideas that worked, it would be interesting to hear about your experiences. I wish I had done this when our daughter was in school.

SHARING - A BROTHER'S TRIBUTE

Details

Written by Ari Geffen

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The following essay was written by Elan Geffen's brother, Ari, for a college class and submitted for this issue of the Newsletter by the boys' mother, Adrienne. What a wonderful tribute to a brother. Elan was born on May 21, 1984 and diagnosed with variant MSUD when he was 20 months old. He has done well.

As a result of attending Symposium '98, Elan was inspired to take his formula by himself. Adrienne adds, "We will make every effort to never miss another Symposium! It was a wonderful experience for Elan to meet and enjoy other kids with the disorder. Besides all the information we received from the professionals, the interaction and sharing with other parents are irreplaceable! My favorite aspect was seeing and hearing the young adults on the panel - VERY INSPIRING."

The person who has influenced me most in my life is a unique individual. He is not a superstar athlete, a revered celebrity, or even a wise elder. He is my younger brother, Elan. Elan is unique because he was born with a genetic metabolic disorder called Maple Syrup Urine Disease. He is one of approximately two hundred* people in the United States with this disease.

My brother has taught me that although life may present many challenges, maintaining a positive attitude can help me ascend the highest peak. Despite the fact that Elan is learning disabled, with mild neurological deficiencies, he maintains many friendships, engages in a variety of athletic and social activities, and volunteers for numerous charitable organizations. Elan's enjoyment and enthusiasm in all of his endeavors are highly contagious, infecting all who participate with him. At baseball, although he does not possess the skills of even a good player, his joy of the game is greater than that of the best players. Every time he gets up to the plate, he comes prepared with his bat and a big smile.

Elan started volunteering at a local geriatric home almost five years ago. He quickly developed friendships with many of the residents, who greet him with kisses, hugs, and handshakes upon his frequent visits. Elan shares his love, and in return they share the knowledge accumulated through their many years.

Elan's life has been challenging. He has had to struggle to compensate for his disabilities. His positive approach has enabled him to achieve substantially more than many have expected. From observing his experiences, I have learned that I can overcome even the most daunting challenge when I arm myself with Elan's contagious smile. Even though Elan may never understand the profound influence he has had upon me, he continues to affect me daily.

***The exact number of persons with MSUD in the United States is not known, however, based on the membership in the MSUD Family Support Group, the number is more than three hundred.**