

# April 1998 OAA Newsletter

## **Kathy's Korner**

*by Kathy Stagni*

Hello again, and welcome to the spring issue of the Organic Acidemia newsletter. I hope this mild winter has helped to keep your children a little healthier this year. I know El Nino certainly helped keep Melissa healthy.

This spring brings some major changes for OAA. In an effort to advance the cause of OAA, the decision has been made to change the organization's board of directors and to add a "parent advisory council".

I am proud to announce that I have assumed the position of Executive Director of OAA and have asked Carolyn Carlson, Menta Pitre and Cindy Winiarski to join the board of directors. These women have been very active in supporting OAA and its mission - Carolyn has been raising funds for research, Menta has launched a parent education and support group for MMA, and Cindy is active in promoting newborn screening and research projects. Carolyn and Cindy live on the East Coast and Menta in the south, adding geographic diversity to the board as well.

The new parent advisory council will help the board by working with new parents of children with metabolic disorders. The goal is to establish a network of parents who will be willing to share their experiences, wisdom and time with those in need of our help. I am pleased to announce that Cay Welch, Deb Harguth, Ilene Osran and Sandy McKillip are charter members of the new parent council. If you are interested in joining please let me know.

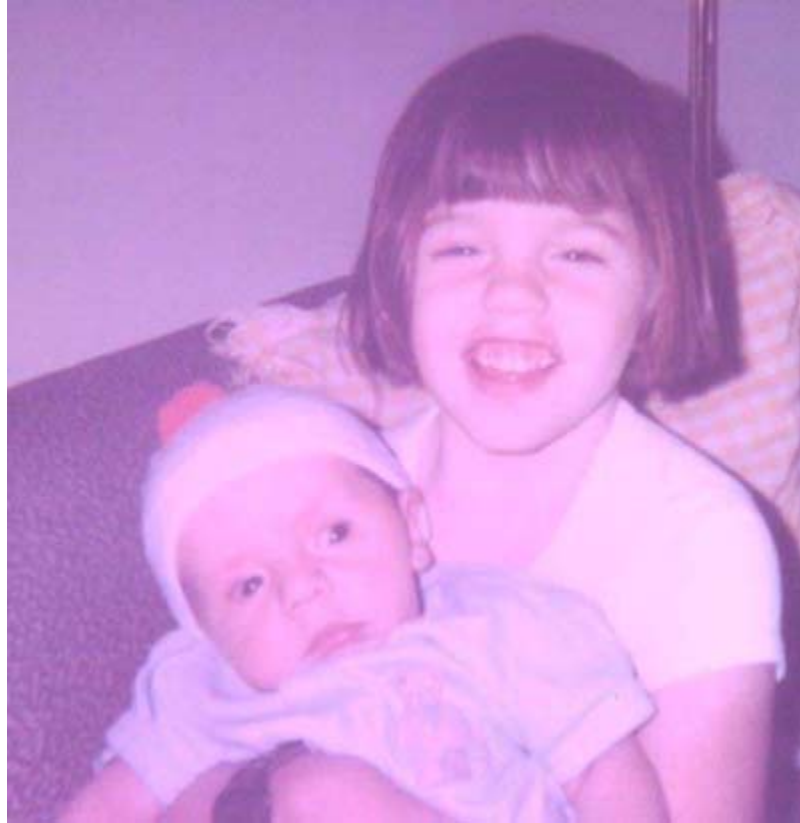
As all of you may know, Carol Barton has been the executive director of OAA for the past 5 years. On behalf of all the parents and professionals associated with OAA, I want to thank Carol for all her work over the past 5 years. She has agreed to continue helping edit the newsletter - and I'm thankful for that as well. I would also like to thank board members Valerie Fulton and Lorie Asten-Neilson for their contributions over their many years of association with our organization.

Your new board and parent advisory council is anxious to begin its mission of providing information, support and visibility for our children's disorder. We want to make this organization more proactive and we need your help. Please let us know how we can better help you!

## Caralee and Jesse Wilkins

### *Methylmalonic Acidemia (MMA), Ages 5 and 12 weeks*

Caralee is one of our greatest joys in life. She is five years old and affected with MMA. My name is Sheila; my husband's name is Duane. We are the parents of four children. Elizabeth is our 10-year-old; Andrew is 9; Caralee is 5 and Jesse, who was recently diagnosed with MMA as well, is now 12 weeks old. Caralee is what her doctor calls moderately affected with methylmalonic aciduria. She has only had a couple of episodes in which she was ill due to acidosis; otherwise, she has been very stable and healthy. Mentally she is slightly delayed, although she is progressing. She socializes well with 2 to 3 year olds or kids that are older (like her brother Andrew's age.) Her speech is significantly delayed. It is that of a two to 3 year old, and so I suspect that is why Caralee



plays well with those who are younger or older. they don't expect a lot of conversation from her. Speech is one of the biggest frustrations in her life (and ours too!). She is just showing interest in books and will sometimes sit long enough to have one read to her. What a delight it is to hear her "read pictures" and sing songs.

Caralee was diagnosed at a year of age due to delayed development and failure to thrive. In July 1997, she had a skin biopsy done. Dr. Rosenblatt of McGill University ran the test. The results showed no missing enzymes that would indicate she has MMA, but in the original labs done at a year old; they showed an elevated level of methylmalonic acid in her blood. The level was actually 11,002 whereas; the high normal range was 271. Her doctor, Susan White of Valley Children's Hospital in Fresno, California says her disorder may be mitochondrial. Has anyone had a similar experience or anyone have a possible explanation? Personally, I feel like I am learning about my children's disease all over again. I wish to be able to identify more closely with other families affected by MMA. It has changed our lives forever.

I mentioned earlier our 12-week-old baby, Jesse. He is healthy, developing at a normal pace. Jesse, like Caralee, had a high amount of methylmalonate acid in his blood at birth. He too, like Caralee, takes carnitine, and B-12. Caralee drinks propimix-2. At this time I still nurse Jesse and as long as his methylmalonate levels stays in the normal range, I can continue. I am amazed by the way organic acidemias can look, appear so different from person to person. I understand this has to do with the enzyme activity that varies in each person. If there is someone who can help me understand this complex disease a

bit better or would just like to write, I'd be forever grateful! At times I feel a little isolated and overwhelmed to say the least!

Thank you Carol and Kathy for your dedication to your families and to all of ours!

God Bless you!

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# Karli Harguth

## *Propionic Acidemia (PA), Age 9*



We are the proud parents of 10 year old Jessica, Karli, (PA) and 6 year old Lucas. Karli was born March 17, 1989. What a nice St. Patrick's Day surprise!

Our pregnancy as well as the delivery went well. Several hours after birth Karli started experiencing apnea and was moved to ICU where she stayed for several days. She was released with an apnea monitor that we named "Clyde". Clyde went with us wherever we went. We were fortunate not to have any more apnea episodes. Karli was taken off the monitor at age 18 months.

We always thought of Karli as a "floppy" baby. She was always very content to lay back and take in the sights rather than crawling or rolling over. She adored her older sister and found her very entertaining. What

need was there to move? Parents are often warned not to compare their children. Jessica walked when she was 9 months old; she was very active while Karli was just the opposite. Karli finally started walking at 15 months and was not very active at all.

Karli was nursed for the first 12 months of her life. We started feeding her solid foods at approximately 8 months. She was a very picky eater, not very interested in eating at all. I remember specifically at 10 months that I felt she was not getting enough protein. For three days straight in February 1990 we worked on increasing her protein amount. I remember how she loved scrambled eggs. After three days she became very irritable, cranky, fussy and not sleeping for naps or at night. We even went out for drives in the car to try to please her but nothing seemed to work. This continued throughout the day and evening. Finally she fell asleep on my stomach late in the evening. During the early morning hours while she was still on my stomach she began to seizure. A frantic call to 911 and she was taken to the hospital by ambulance. While in the hospital she had several more seizures. To make a long story short, there was a neurological doctor on staff who recognized Karli's low muscle tone as not being normal. She then ordered several tests to be run on both mom and baby. After a month of testing and waiting Karli was diagnosed with PA. We later learned that Karli does have a small amount of enzyme activity which is why she was not diagnosed until 10 months of age. We were referred to University of Wisconsin's Children's Hospital in Madison, Wisconsin. We were under the care of Dr. Wolff until Karli was 4 years old. We then transferred to Mayo Clinic in Rochester. Karli was in and out of the hospital numerous times in her early years of life. We have managed to be hospital free since 1995.

During Karli's early years it was a struggle to manage her diet. Between restricted protein levels and correct calorie numbers it was hard to regulate the diet. There were many midnight calls to doctors and ambulance trips to the hospital. Karli has been a trooper through all this. It is amazing how well the kids can handle this; it is the parents that go nuts! As Karli has become more responsible for eating and drinking her

formula (which we call bug juice), which has made it easier to manage her diet. Currently Karli is on a 32-gram protein diet; her daily intake consists of 32 ounces of formula made with 80056 and Maxamaid, 17.5 ml of carnitine, 480 mg of L-Valine and 200 mg of L-Isoleucine. Karli is a third grader at Caledonia Public School and is doing well. We pack her "lunch" daily, she has had the pleasure of eating a few school lunches this year (hot-dog and chicken nugget days). When she eats school lunches we balance the day by lowering her protein intake at supper. This doesn't seem to bother her because she is thrilled to be eating lunches like the other children.

Our visits to Mayo Clinic have become fewer in the past years. We now see our wonderful Dr. Gerrald Vockley 3 times a year. We also get blood work done between our visits to at our local clinic. The blood work is then sent to Rochester for analysis. Karli has her older sister, Jessica and younger brother, Lucas to keep her busy and active in several activities. Karli enjoys typical third grade activities. She has a harder time keeping up or doing as well but she tries her best. She does struggle with being physically active. When running, riding bike or sledding she is only able to do so much activity. When she over exerts herself she ends up gagging and throwing up.

Life with Karli certainly has had its ups and downs. When Karli becomes sick, life is put on hold until we get back to normal (or negative ketones in her case). This is sometimes difficult for our other two children to understand. We have a wonderful "extended family" that has always been there to lend us a hand to help us through the down times. We are very thankful to have them. We also have a great school staff to work with; they have been there to help us out with Karli's academic work. The medical staff that surrounds us has always gone that extra mile to keep Karli on an even keel. Also the people we have met through our dealings with PA, especially the Stagni's have been wonderful. It gives families great support to be able to get together and share problems and advice. We truly have been blessed in many ways.

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## Nutrition News

### *Energy and Protein Requirements of Infants and Children with Inherited Metabolic Disorders*

(Taken from the *Metabolic Currents*, authored by Phyllis B. Acosta, DrPh, Rd,  
Director of Metabolic Research and Development at Ross Laboratories

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Infants and children with Inherited Metabolic Disorders (IMD's) have the same growth potential as normal infants and children. Infants with IMD's, given adequate energy and protein, based on 50th percentile weight-for-age grow normally. Energy and protein intakes, prescribed on the basis of actual weight in the failure-to-thrive infant, with no allowance for catch-up growth, lead to growth that drops further and further below normal. For catch-up growth to occur, energy and protein intakes should be based on 50th percentile weight-for-age. Energy and protein requirements may be greater than normal when L-amino acids are the source of protein equivalent.

#### Weight, Energy, Protein Requirements

Category Age (Years)	Weight (Kg)	Energy (kcal/kg)	Protein (g/kg)
Infants 0<3 mo	4.6	120	3.00-3.50
Infants 3<6 mo	7	115	3.00-3.50
Infants 6<9 mo	8.6	110	2.50-3.00
Infants 9<12 mo	9.7	105	2.50-3.00
Children 1<2	11.5	106	2.05
Children 2<3	13.7	103	1.92
Children 3<4	15.8	97	1.82
Children 4<5	18.8	94	1.77
Children 5<6	19.5	90	1.70
Children 6<7	21.8	86	1.68
Children 7<8	24.0	80	1.68
Children 8<9	26.5	73	1.68
Children 10<11	33.0	73	1.67

*(Editor's note: This information is provided as reference since protein intake is a common challenge for managing children with metabolic disorders. The summary chart outlines the energy and protein levels at various ages/weight intervals. This information is not a substitute for following the direction of your dietician and/or physician who must consider many other factors when determining the optimum protein intake for each child.)*

# Inborn Errors of Metabolism Diagnosis and Treatment

**Emmanuel Shapira, M.D., Ph.D., Professor of Pediatrics and Biochemistry**

**Eva Kozicz, M.D., Neonatal/Genetics Fellow**

**Tulane Medical Center, New Orleans, Louisiana**

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Inborn errors of metabolism are individually rare, although cumulatively occur in about 1 in 5000 newborns. Most of them are inherited as autosomal recessive disorders. This means that both parents are obligate carriers and the risk of recurrence is as high as 1 in 4 in a future pregnancy. Those inherited diseases of X-linked recessive fashion have a risk of recurrence of 1:2 in male offspring with 1:2 risk of carrier status in female offspring. The carrier daughter may partially express the disorder. Prenatal diagnosis is available for most of these diseases, if the diagnosis was confirmed in the index patient.

Most inborn errors of metabolism present with nonspecific symptoms like an acute episode of infection, or loss of consciousness with or without seizures or vomiting. Frequently they have an acute, severe onset and a recurrent character. Inborn errors of metabolism were found in several patients previously diagnosed with near - SIDS (sudden infant death syndrome). Another possible presentation of metabolic disorders is failure to thrive in an infant with or without developmental delay or seizures. Even the same disease can express in different family members with different presentations. Marked variability exists in the age of onset (neonatal - to late infancy). Very many of these disorders are amendable to an effective treatment which prevent the sequelae of the disease.

The initial work up should be done early in order to rule out the possible inborn error of metabolism. This should consist of the blood tests (glucose, acid-base status, electrolytes, lactate, ammonia, blood count and liver function studies) and urinalysis. If there is a high suspicion for metabolic disease, serum amino acid and urine organic acid and carnitine analysis is recommended during the acute episode.

The treatment of patients with inborn errors of metabolism includes restriction of dietary components leading to the clinical manifestations and supplementation of one of the cofactors in pharmacological doses and in some of the metabolic diseases the use of alternate pathways to the metabolic block. In many patients, especially those who have disorders of inborn errors of organic acids, carnitine therapy is required. Most of the patients benefit from this treatment and in some of them all of the sequelae of the inborn error of metabolism could be prevented.

In many of the metabolic diseases acute exacerbations do occur as an outcome of an intercurrent infection. In some of these this could be a life-threatening episode. Patients should be evaluated by a physician and treatment and management prescribed by telephone conversation should be avoided. Acute exacerbations often require discontinuation of all feeding, intravenous administration of fluids, electrolytes, and glucose in an attempt to correct hyperammonemia, hypoglycemia and the changes in acid-base balance.

The long-term outcome for most of these metabolic diseases is improving as a result of early detection and an effective treatment and management in long term treatment and in the acute exacerbations.

The role of parents and family members in loving care and stimulation, occupational and speech therapy and special education are extremely important in these children in order to maximize their potential. Support groups can help the families with information and friendship to cope with their several difficulties in managing a child with metabolic disease.

# Michael John Dalton

## *Methylmalonic Acidemia (MMA), Age 9*



Michael, born 5/16/88 is almost 10 years old and I have always wanted to write something to put in this newsletter, but I just never finished writing. I have a hard time remembering everything that has happened to him since he was born. I told this to Kathy, and she said just write about how he is doing now. So here I go. Michael was diagnosed at the age of 8 months after 4 months of knowing that something was wrong. Finally, we had an answer, of course not the answer that we wanted, but at least he hopefully would get better. Then we waited another couple of months for his fibroblast (skin cell culture analysis) to come back. He is B-12 responsive, type Cobalimin C.

When Michael was younger he seemed to be sick much more than he is now. He sure has been through his share of hospital stays! Michael also had a stroke in 1994 and still does not have full use of his left hand. He also has seizures, which currently are pretty well controlled; he is on Phenobarbital and Dilantin. Michael also is visually impaired but is getting better each year.

Currently he is about the age level of a 12-18 month old. He is a very loving boy, full of laughter and enjoys the simplest things in life. He loves music; Barney and Raffi are his favorites. Michael also loves to bug his younger sister Ashley (5 years old). I'm surprised that she still has hair left on her head! He also loves to wrestle! He loves the sound of laughter, but is very sensitive when he hears anyone cry (even if it's on a TV show), then Michael starts to cry too. Michael loves school, he is in the 4th grade MSMI (moderate-severe mentally impaired) program. Michael does not talk or use sign language. He does say "ma-ma" which of course makes me happy.

He loves to eat; of course this is difficult when he is on a restricted diet. He gets 1.5 grams of protein per kilo, so as he gets bigger it is easier. Michael weighs about 50 pounds and seems to be getting taller and taller every day. He is a challenge, because he loves to get into everything and anything he can get his hands on!

We love hearing from other parents and this newsletter is a great help to us. Thank you!

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# Chad Schuffler

## *Propionic Acidemia (PA), Age 5*



Writing an update on our kids is exciting and challenging, just like taking care of them. I am never sure where to begin or how to organize the message, and Lord knows it is never ending! Let me begin by saying that the first time I wrote about Chad was just before he turned one year, and at that time I had no idea how his story would unfold. For that matter I still don't know what tomorrow will bring, but I now have a better idea. Chad will be FIVE years old this May! I truly can't believe it and neither can his doctors. From the pediatrician to the geneticist to the surgeon, they can't believe Chad and his strength and will to fight.

Beginning in November of '97 we started Chad on TPN (total parenteral nutrition or IV feedings) because he wasn't tolerating his full feeds (Chad is fed by G-tube exclusively). Chad was hospitalized in November, '97 for an endoscopy to see if there was any problems with his git (gastro intestinal tube). The only thing found was that his stomach lining seemed poorly nourished, probably due to his low tolerance of protein. Chad will only tolerate

about one gram of protein per kg of his total body weight via "oral feeds" (via g-tube) making his total protein intake from oral feeds in 24 hours about 14 grams. So, after contacting Dr. Steve Kahler, who is experienced with TPN therapy for PA patients, we started Chad on TPN. Chad was to receive TPN every night for 10-12 hours for 6-8 weeks. After that period of time, his stomach should have rested and his stomach lining should have been nourished, as he would have received the extra protein via the TPN. Well, after the 8 weeks I weaned him off the overnight TPN and to our dismay Chad started vomiting again. For whatever reason Chad just does not tolerate large volumes of formula. To this day we are still running TPN every night.

Having to run TPN on a regular basis requires quite a bit of extra work. Chad has a hickman catheter, (central IV line going directly to his heart vessels), which requires sterile dressing changes regularly, and he is not allowed to go in public pools or the ocean (a big bummer when you live on an island). I have to check his labs about 3 times a week, and of course there is the regular communication with doctors, nurses and the pharmacy that supplies us with all the necessities required for having a central line and running TPN daily. BUT, even though it is a lot of extra work and the constant threat of central line infection, which can be life threatening, Chad is doing remarkably well. In fact Chad is progressing faster and staying stable longer than he ever has. Chad finally has a full head of hair, fingernails, and is gaining weight on a regular basis. To some this may seem trivial, but for us it is a milestone. Until we started the TPN therapy Chad was always sick. At least once a month he required continuous, 24 hour, IV therapy, for acidosis and hyperammonia, his progress was minimal and if he gained weight he would just lose it with each episode of acidosis.

So, while TPN is questionable for PA patients and expensive (I understand the cost of the TPN that we use is about \$100.00 per bag, which is \$100.00 per day as we use one bag every night). So far we have it

approved through Medicaid, as he is classified as 100% disabled. I might add that he does receive SSI. We are not using PA specific TPN because of the high cost of that type (which is about \$1K/bag), not to mention the availability. The TPN is a "home brew" - a general mix minus the lipids and that is why it is only one half gram per kg and why we must keep an eye on his blood serum amino acids, so that we not have overload him with MVTI, (methionine, valine, theronine and isoleucine). Having a central line is not the greatest thing to have; it is all-ok for us, because it is working!

One last thing, our thanks to Dr. Wanda Meurs, Dr. Hsia, Dr. Kahler, Dr. Powell, Pharm-a-care, and our hospital lab for all your help and support.

Until next time God bless each of you and don't ever give up. You can reach us at:

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# Newborn Screening for Organic Acidemias

*by Stephen Kahler, MD*

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*(Editor's note: Most children with organic acidemias are diagnosed after one or more episodes of catastrophic illness or vomiting and poor growth. Recent developments in newborn screening now offer the prospect of early diagnosis for all babies, leading to earlier treatment and improved outcome. We feel fortunate to have Dr. Stephen Kahler write us this article on this improved newborn screening procedure.)*

Screening of infants for metabolic disorders began in the 1960s in the United States and Europe. The first screening programs were for PKU (phenylketonuria). Several discoveries and inventions were essential for the success of these programs, including: 1) the disorder was treatable; 2) the testing was relatively inexpensive; 3) the disease could be diagnosed before damage occurred; 4) the test could be done with minimal inconvenience to the babies and their families, since relatively few infants would actually have the disease. Bob Guthrie and others developed the method of applying blood to filter paper cards, letting it dry, and sending the card to the laboratory. The original Guthrie test for PKU relied on a microbe that cannot grow unless there is excess phenylalanine in the blood spot. John Hill and George Summer developed another test, using a chemical method to analyze phenylalanine, in North Carolina. Since the 1960s many other tests using the Guthrie cards have been developed, including tests for hypothyroidism (if untreated in infancy this can lead to a form of mental retardation called cretinism), sickle cell disease and other disorders of hemoglobin, congenital adrenal hyperplasia, galactosemia, maple syrup urine disease, homocystinuria, and other disorders of amino acid metabolism.

Organic acidemias typically don't lead to diagnostic changes in amino acid levels, and urine organic acid analysis is not suited for mass screening. The discovery of acylcarnitines by David Millington, Charlie Roe, and their colleagues revealed a whole class of compounds which reflect impaired organic acid metabolism present in many of these disorders. The analysis was in sophisticated instrument called a mass spectrometer. The initial technique used urine, from a patient who had been given carnitine, and was suitable for testing individual samples from sick patients. The subsequent development of analysis by tandem mass spectrometry (tandem MS) allowed us to use blood, without giving carnitine first. The new technique needs only a very simple preparation of the plasma or blood spot sample before rapid analysis in the instrument. This test has been used for diagnosis of sick patients and testing their relatives for the past decade. Most of the development of this test was done at Duke University, where I worked from 1983 to 1997.

Major changes in how the test is done have simplified the process. These improvements include sample preparation using robotic devices, automated sample injection into the instrument, and computer-assisted interpretation of results. Newborn screening using blood spot samples and tandem mass spectrometry is now being implemented in developed countries around the world. Acylcarnitine analysis will identify over a dozen disorders, most with extremely high reliability. The most common of these disorders is MCAD deficiency, which occurs at a frequency of 1/10,000 to 1/20,000 or so in most populations derived from Northern Europe. Diagnosis is virtually 100%. Isovaleric, propionic, and methylmalonic acidemias are readily recognized, as are most cases of Glutaric aciduria type I. The fatty acid oxidation disorders VLCAD/LCAD, LCHAD, CPT I, and CPT II deficiencies, and the mixed disorder GA II deficiency are also diagnosable in most cases. All these disorders are apparent from one test.

The same sample can also be used for detecting amino acids by changing what the instrument is looking for, during the analysis--a second test is not necessary. Phenylalanine, tyrosine, leucine plus isoleucine (for

MSUD), and methionine are some of those detected. Thus the tandem MS test can replace the existing testing done for PKU and other amino acids by older methods.

The tandem mass spectrometer is an expensive instrument (typically a few hundred thousand dollars, but prices are coming down). Even though most of the disorders are rare, by being able to test for many disorders at once the cost per diagnosis is reasonable. Many of the organic acidurias (but not usually MCAD deficiency) can lead to severe illness soon after birth, so rapid testing is essential. Even if the child is sick before the newborn test is completed, knowing the diagnosis quickly (from a sample obtained while the child was still well) can speed up appropriate treatment. A screening lab should be able to complete the test within a day after the sample arrives. The results of the new testing programs should include faster and more accurate diagnosis of organic acidemias and related disorders, so that infants will be identified before catastrophic illness has begun, or else earlier in the illness so that better treatment can be started sooner.

Much of the work to develop newborn screening using tandem mass spectrometry was done at Duke University, particularly by David Millington, Don Chace, and Steve Hillman, with support from the state of North Carolina. The method has been adapted and placed in service in a private laboratory offering screening tests to supplement existing state-sponsored tests (Dr. Ed Naylor at NeoGen, Inc., in Pittsburgh). Many hospitals are now sending samples to two labs. North Carolina is the first state to embark on statewide screening (on a pilot basis) using tandem MS, in collaboration with NeoGen. Other states and regional programs are seriously considering establishing tandem MS as part of their newborn screening programs.

Newborn screening using tandem MS is also being established in several centers in Great Britain. In my new home of Australia three states (there are 5 screening programs here, for the 7 regions) are establishing similar programs, and many centers in Europe are intending to do likewise.

In addition to amino acids and acylcarnitines, several other types of analysis are possible using these instruments. It is their versatility that makes them so attractive, for once the instrument is in the laboratory the operating costs are minimal, the precision is very high, and the need for repeat testing is very low. I expect many other tests will be added to acylcarnitines and amino acids, and tandem MS will be the main tool for much newborn screening for the foreseeable future.

## **Kristy Hare, Propionic Acidemia, Age 33**

*(Kristy wrote me a note this past Christmas, and I asked her permission to reprint it in the newsletter)*

Not much to tell as far as my metabolic disorder is concerned. I am still on a low protein diet (which is easy for me because I do not like most meat and do not believe in eating animals much anyway). I got the flu shot this year, and was only knocked down with it for about two days versus two weeks. If I get overly weak or tired, I up my sugar intake. I am not on any certain medications except for migraine headaches. I am taking Inderal as a preventative and Imatrex (the injection) when one breaks through. Doctors told me that my migraines are due to the heredity and hormones. Certain food triggers (in my case, yogurt and peanut butter) and stress also affects them. I've always been curious though if other people with Propionic Acidemia suffer from migraines often. Perhaps, genetics may have something to do with them.

On the home front, things are stable. I have a new job as a switchboard operator for a company that manufacturer treadmills. I love it! The company has a bible study and a fitness room, which I am involved in. I am dating a nice guy now and attending Junior College, taking one class at a time toward my AA. I just finished a course in child psychology, which was the toughest course I've taken, and surprised myself by passing it with a "C". I am a member of PETA (People For The Ethical Treatment of Animals) and write letters to everyone supporting animal rights. But I won't go overboard by rallying at demonstrations or ruining fur coats, which I'm sure you've heard about. I am also the Secretary of our Homeowners Association, which is rather exciting since I am the owner of a condo.

The cats are all fine and still, very spoiled. Unfortunately I don't have a picture to send now. I've been mailing flood care packages of victims all over the states—hope all is well with you and your family.

**Kristy Hare  
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## **Cadence Pierce, Propionic Acidemia, Age 18 months**

Hey Everyone:

We hope everyone had a great holiday season. There have been some big changes in the Pierce household. A week before Thanksgiving Cadence (18 months) had her G-tube put in. The NG she had been using for the prior four months was creating more problems than it solved, so we decided to take the plunge. We worried, of course, but we felt as if we were backed into a corner.

She came through the surgery with flying colors. The GI surgeon was surprised how well she looked inside, he said she was perfect internally and the surgery went well enough to put in a textbook!

The day of the surgery she stayed knocked out on Morphine because she was so sore, but by two days post-op she was all over the pediatric floor. Two weeks later she got her button put in. Life is so much easier now.



On top of all the blessings that have come our way lately, Cadence took her first steps on December 28. Another good thing that has come of all of this is that I am back in school yet again. I graduated with an Associates in Science and an Associates in Arts a month before Cadence was born with the intention of getting my Bachelor's in Chemistry. Needless to say, life after Cadence's birth was tumultuous and my previous plans got postponed. Now, because of the large part of our lives that is consumed with the medical world, I decided to go to Nursing school. I may as well get paid for what I've had to learn over the last 18 months. School's been tough, but it will be worth it in the end, and Cadence benefits even more. I'm very fortunate that the college I attend is only five minutes away and one of the best nursing schools in the state. Cadence's aunt is available to watch her while I am in school, which is very good since she provides more attention to her than a day care could.

Easter Seals discontinued Cadence from her regular speech therapy because she is over her oral defensiveness (but I say it's because she talks ALL THE TIME).

Overall, Cadence is happy all the time and laughs a lot! She certainly makes us laugh a lot. She continues to remain ahead of her age cognitively and a little delayed with her gross motor skills, but maybe that gap will close some now that she's figured out walking. And as you can see from the picture, she's living up to her name!

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## Letter from Cay Welch

Dear Kathy:

Welcome to the new year. IOGA, International Organization of Glutaric Acidemia has been busy meeting and hearing from GA families.

Our mission remains the same - to further prevent neurological insult of Glutaric Acidemia children. Prevention via education and prevention, advocating of the newborn screening test, research on metabolic disorders and distribution of GA booklets to doctors for diagnostic purposes.

Metabolic research on Glutaric Acidemia is taking place presently at Baylor Research Institute. IOGA granted monies in 1997 for a metabolic clinical trial study. We await the publication and results of this study.

As for education, 68% of all births in our home state, Pennsylvania are screened for metabolic disorders via the newborn screening. This is largely due to the collective efforts of Dr. Holmes Morton and Dr. Edward Naylor. To date, Dr. Morton treats approximately 50 Glutaric Acidemia children at his clinic for Special Children in Lancaster, PA. Dr. Edward Naylor is director of and president of Neo Gen Screening, Inc., a private laboratory doing the newborn screenings. Dr. Naylor recently returned from Mexico City where he was asked to consult on a potential screening laboratory site. Dr. Naylor provides assistance in the initial set up and training on the necessary tandem mass spectrometer and laboratory equipment. We included his report in our IOGA's last newsletter regarding the hospitals on board doing the testing.

*(Editor's note: If you would like to see a list of hospitals, please contact me.)*

IOGA was successful in 1997 in raising \$30,000 by having two different fundraisers plus a successful grant approval. Our first fund raiser took place August 6, 1997 where we had an auction, raffle, food and entertainment. In December we had our Baskets of Love benefit auction tied into a Xmas auction with a local auctioneer.

One of our objectives in 1998 is to sponsor a graduate student to do a PHD on Glutaric Acidemia and its impact on brain injury. We would also like to distribute the GA booklet to medical teaching facilities, pediatric practices, specialists and family physicians.

In January, I testified at the State of Massachusetts Public Health Laboratory regarding the expansion of the Newborn Screening test. The New England Regional Board is like no other location in the United States. It is the only place we know of that control the screening test in 6 states. The test, as it stands now, only screens for only 8-9 disorders. They are considering expansion to test of treatable metabolic disorders. In Massachusetts, they charge \$57 for screening of 8 disorders. In Pennsylvania, Neo Gen, run by Dr. Naylor tests for 31 treatable disorders, including Glutaric Acidemia. Specifically they are looking for parents to testify whose children were damaged by not having the screening done. From the sounds of things, we collectively had an impact on the decision making for the expanded screening. IOGA patiently awaits the outcome of results from the New England Regional Boards decision. Every child screened and parent educated on this most treatable disorder is a success story.

Next up on our calendar will be March 28, the Metabolic Conference. If you are interested in our newsletter or fundraising, please feel free to contact us.

To Better Health!

**Cay Welch**  
**International Organization for Glutaric Acidemia**

## **Congratulations Carol!**

"After knowing him for 10 years at church, and dating since the new years dance, I am pleased to announce that I am planning to marry," reports Carol Barton.

While no date has been set yet, Carol says that her beau is Frank Charles Owens III. He has a son, Frank IV, who is a month younger than Carol's daughter Bethie is. He also has a daughter, Danielle, who is a year older than James, but in the same grade. "Like me, Frank likes camping, going to church, the color blue, and we even have the same pediatrician at Richmond Kaiser," she said. "I am enjoying myself and being in his company. Amazingly I feel like a teenager again. It's nice to be in love!"

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## **Special Thanks**

The OAA exists with the generous contributions of both parents and corporate sponsors. As costs rise, and as we expand our programs, fundraising becomes a very important element of our future. We appreciate the many gifts that all of you have made to keep OAA growing strong. We are especially grateful to those who have made financial contributions beyond the annual newsletter subscription fee - thank you.

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## **OAA Web News**

OAA's new web site has received over 900 hits in one month! Ninety percent of the calls I receive are from parents or professionals that heard about OAA on the Internet. Our web site is hosted by Comtrol Corp., a St. Paul, MN based computer products manufacturer. I would like to thank them and their technical staff for their continuing support and generous donation of time.