

## Editorial

*by Kathy Stagni*

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I'm sure everyone of you have said – "whew, where has the time gone" — well that's exactly how I feel. I can't believe it's June already. I love this time of the year, the weather is good and our children tend to stay healthier during the warmer months.

I've been busy since the last newsletter went out with the large amount of donations that have arrived for the Organic Acidemia Association. I wish I could print each and every one of your names in the newsletter – but I'm afraid I will fill up an entire page! Thank you so much for your annual donations that have arrived since the January newsletter. I have not focused on fundraising since I took over OAA five years ago. It's not one of my favorite things to do. Although, I must admit, I have started to fundraise for the Propionic Acidemia Foundation and I was pleasantly surprised how many of our friends and families were happy to give to charity that might help find a cure or improve the life of our Melissa. Most people do give to charities throughout the year – so it might as well be to a charity that will help the lives of someone they know. I truly believe that staying busy and active in something that you are passionate about while taking care of a child with a chronic disability is the only way that you stay "sane". OK, maybe there are some days that I'm "saner" than others!

Thank you to all that have contributed stories and articles for this issue of the newsletter. We are blessed to have a nurse practitioner, Lynne Wolfe write some articles that should be of interest to many of you. After reading how parents were interested in learning about osmolality and antibiotics on the OAA listserv, I asked Lynne (who is also a OAA listserv member and her contributions are greatly appreciated!) to write the articles for the newsletter. Thank you so much Lynne! After thirteen years of dealing with metabolic disorders I was surprised that I had never heard the word "osmolality"....so it's refreshing to have her explanation in this issue of the newsletter. On the other hand, I hope the article written by Dr. Olaf Bodamer isn't too technical. That's why I've asked Dan Osran, dad to Jessica, MMA, Cbl C to write a parent's perspective summary. If you're like me, it may take many, many times reading about these disorders before something can really sink in!

I've also included the registration form for the upcoming National Coalition for PKU & Allied Disorders Metabolic Conference being held in Orlando, Florida on Thursday, October 3rd. We are so fortunate to join with the World Congress on Disabilities Conference, which is being held at the same location and time as our conference. A family reception will be held at the hotel the evening before the conference, October 2nd. We also hope to have an afternoon/evening set aside where we can plan a family day at Disney World. I want to thank moms, Dena Royal, Doreen Dix, Lori Sanchez, Melodie Jones, Galen Welch, Karen Dalton and Michelle Pfingstag for volunteering to help. These parents will assist me in making this conference a rewarding experience for you and your family! See you in Orlando!

# Jennifer Mabbott

*MMA, Mut 0, Age 5*

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Hi Everyone,

It's been three years since our first story in the newsletter, so I thought I'd give you all an update. My daughter Jennifer is five years old and has MMA mut0. Our introductory story was in the January 1999 issue of the OAA newsletter.

We've come a long way since that letter and have had many ups and downs. When Jen was 27 months old, she had a preschool assessment done. Her overall development was equivalent to a 17-month-old, which translated to a severe delay in all fields. But that's not a bad thing...it's only numbers and being at a severe level enabled us to be eligible for various services.

We did an 8-week program at Glenrose Rehab Hospital. The children got to interact in a playgroup setting with all the various therapists present: speech, physio, occupational and psychology, all observing and helping. Jennifer and I both enjoyed it. In June of that year, Jen had six weeks of speech therapy, one hour a week and it helped wonderfully. Jennifer FINALLY started talking, and I don't think she has shut up since!!

The following September she entered the Junior Nursery program at Glenrose Rehab Hospital. This was a program where the children get their individual therapies where needed most, plus they had the social interaction with their peers. The parents were required to stay with their children, but once a month we had Parent Group, where we got to chat and offer support and suggestions with the other parents. It was a nice welcome break.

This same school year, we also were enrolled with a playgroup through Early Intervention. It was much along the same lines as Jr. Nursery, but not as much therapy and not as structured. Jennifer enjoyed this more as she got to run and play as SHE wanted! So both of these programs were of great help and Jennifer progressed nicely. She graduated that June from Jr. Nursery, with an assessment that showed her improvement. Unfortunately, she had improved so much, that we no longer qualified for any of the programs through Glenrose.

In March of 2000, we were very fortunate to attend the New England Conference in Boston. Unfortunately Jennifer ended up getting really sick, so that put a damper on things, but all in all it was a great opportunity to finally put faces to some of the names I see floating around so often. It was very educational and reassuring to hear of others stories that were so similar to ours! Turns out we aren't so weird after all!! Jennifer ended up being much better by Sunday, so we were able to do a little sightseeing in Boston, so I was still very grateful that we got to attend this.

Halloween of that year, we found out Jennifer has an allergy to peanuts. One little Reese's pieces in her Halloween goodies confirmed that! But it wasn't an acute reaction, so we were lucky and have just been extra careful reading labels now! It seemed that about every couple of months, Jennifer would have a metabolic upset, but they were nothing terrible, just lethargy, no appetite, usually vomiting, but I would adjust her formula, take out the protein and double the Carnitine and usually within 24 hours she was back to herself. We've only had about five or six ER visits with only four of them ending up in admissions. Of course it was always on a holiday or a long weekend, I'm not sure how that always happens

that way!!! It seems the year 2000, she had a lot of respiratory problems. At Easter she had bronchial pneumonia and in June she ended up with R.S.V. Then Boxing Day that year she was diagnosed with Asthma! WOW! Another thing for me to learn about and watch out for! So she now has a puffer that she is required to take twice daily and we seem to be keeping things under control.

We had just gotten the Asthma thing under control, when at the end of January, we had a bout of the flu go through our house and of course Jennifer got it. After 13 hours of vomiting any fluid that she would take, I knew we were heading for trouble. It's really hard to explain but Jennifer was just "different" this time. She had an ungodly wail that still sends shivers up my spine when I think about it, and by the time we got to the hospital, which was about 24 hours after this all started, she was semi-comatose! Her ammonia level was 208 and we had the whole trauma unit in an uproar. They thought she was having a stroke at first, but a C.T. scan confirmed that there was no fluid or swelling of the brain. They ended up sending Jennifer to the PICU and she remained in a coma for about 20 hours. Jennifer would usually crash fast, but would bounce back just as fast when treated. We ended up going home after five days in the hospital, but it was not with the same girl that I had a week before! Jennifer had to learn to do everything all over again. She had to learn to sit up, walk, talk, just about all of her motor skills. It was very frustrating for her and it was extremely heart wrenching for us watching her. A MRI later on did confirm some minimal brain damage, but the doctors don't feel that it is irreparable. I'm happy to say that she is coming along well. She cradles her one arm when she walks, and also drags her one leg. We have just passed the year mark of that horrid bout and I am now breathing a little easier.

As far as eating, well....Jennifer still has her G-tube, but I would not be without it! It has been a godsend! It makes life so much easier, especially when she's in a crisis. Since her bad episode last year, we had a major change in her formula, and she no longer takes any by mouth. We also no longer use the overnight pump. We found she was moving around too much and too many times we'd go to check on her, only to find the tube around her neck. So I bolus all of her formula throughout the day. Her formula consists of Propimix 2, Prophree, Enfalac with iron, skim milk powder and her Carnitine. Her appetite is improving as she grows older and when in the mood she is pretty good at trying just about anything. She tends to prefer salty over sweet though. She's not big on candies, but does indulge in the occasional chocolate. Her favorite foods are chips, cheezies, chips, popcorn, chips, instant noodles, chips, the wraps and broth from won ton soup, mac and cheese, mashed potatoes, oh yeah...and did I mention chips???? She loves McDonald's fries and gravy, will nibble on a piece of just about any meat, and will occasionally have a drink of "our" milk, but her one old standby is still her baby pablum. She still has to have her pablum, but it's a help in getting all her daily formula into her and I add applesauce to it as well and I guess it is healthy! She is gaining weight like crazy, I think she is now 45 lbs., and is getting to be quite a "load" to carry!!

Jennifer has accomplished a lot over the past three years. A MAJOR accomplishment this year was potty training!!!! YES!!!! We are finally going on the potty. The day after she turned 5, she said "I'm 5 years old now, I don't need to wear diapers!!" And guess what? Other than at night and when we go on long trips, she doesn't wear diapers anymore!

I didn't put Jennifer in kindergarten this year. She could have gone, but seeing as how her birthday is in November and she's delayed a bit anyhow, why try rushing things? She really got quite "clingy" after her bad episode last January, and she's just starting to be comfortable going to other people.

We are now living with Jennifer's dad out on a farm! She is definitely daddy's little girl. She has him wrapped pretty tight and oh, does she know it! She can work that pretty good!! Jen just loves being out in the country with all the room to run and all of our animals (cats, dogs, sheep, ponies and a donkey). I think Jennifer is a lot happier and more content with her dad here and I think that has a very positive effect on her whole being. It's also nice for me to have someone to share the burdens and the worries that this disorder tends to bring with it at times.

All in all, things have been going pretty well. I am much more comfortable and relaxed with Jennifer and her MMA. I still worry when she's sick, and occasionally I wonder what is going to happen, but I try not to dwell on the negative. I take each day as it comes. My new motto is this: Yesterday was the past, tomorrow's the future, but today is the present, so treat it as the gift it was so intended to be. God has blessed us with these "special" children and I think these kids have some pretty "special" parents. The OAA and its network of families are still my support and backbone. I've actually been the one giving advice and offering support to some of the new families now! I'm scared to even think of where I'd be without all of you. Jennifer has changed our life enormously. I couldn't imagine life without my Jen. You are all in our thoughts and prayers every day. God Bless and take care!

We'd still love to hear from any of you, just to chat or with any questions about what I've written about here.

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# What is Osmolality and Why do we Care?

*by Lynne A. Wolfe, MS, PNP, BC*

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Osmolality is a chemical term used to describe the number of particles in a certain amount of water. The higher the number of particles in relation to the volume of water, the more concentrated (or, hypertonic) body fluids become; the lower the number of particles to the volume of water, the less concentrated (hypotonic) body fluids become. The human body functions best with an Osmolality of ~285mOsm; we call that Isotonic. Children under the age of two years old have a normally higher percentage of total body water. Their body water is distributed to different places than an adult's. They use up and need more water daily by body weight than adults, and are more sensitive to changes in the Osmolality of their blood than adults. Adding to the difficulties, children under the age of two years old also have immature kidneys; that is, they have difficulty getting rid of too much water, and also don't conserve water when it is needed by the body. Osmolality can change rapidly with extra fluid loss from fever, fast breathing, sweating, vomiting, or diarrhea, if you drink too much or too little water, or if you are losing extra water through the kidneys.

In children with OAs, several more things can create difficulties for them to maintain a normal Osmolality. First, many of them lose more salt and sometimes proteins, in their urine than is normal. Whenever salt and proteins leave the body they are diluted in extra water. Metabolic formulas can potentially have high Osmolalities, because the number of particles is high in relation to the amount of water. In order to restrict certain amino acids, all proteins must be broken down into amino acids, then certain amino acids removed depending on the OA being treated. By the definition above, a whole protein is one particle versus the many particles made by single Amino acids. These many particles are called the Osmotic load. If Metabolic formulas are too concentrated; abdominal cramping, nausea, vomiting, diarrhea or constipation, can occur, associated with irritability and/or lethargy. If not corrected, weight loss or poor growth can occur. In children with OAs, nausea and vomiting may increase the risk of aspiration, diarrhea can cause severe diaper rashes and ongoing nutrient loss, and weight loss usually triggers muscle breakdown leading to increased Amino acids in the bloodstream causing metabolic instability.

When your Dietitian is calculating the calories, proteins, and certain amino acid amounts for your child, they also calculate "free water" needs. In certain situations, your Doctor may need to order extra free water. Your child may need extra water, with salt added (Pedialyte or Normal Saline), to make up for extra water losses from fever, respiratory illnesses, vomiting or diarrhea, or excessive sweating. Water will follow salt, so when we use Pedialyte or IV fluids, we are giving water and salt, and many times sugar too. The extra sugar helps stop the muscle breakdown by providing an easier source of energy for our body's cells.

## References:

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# We Come From the Land Down Under

*Rachael Sharman*

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OAA listserv enthusiasts will be familiar with me as the regular Aussie contributor "Stuart Mackintosh" who always curiously signs himself as "Rachael, Mum to Sarah 3-MCC!" What you may not know is that I am the Vice President of an Australian group, the Metabolic Dietary Disorders Association. The MDDA was set up to help any person or their family deal with the complex management of inborn errors of metabolism requiring dietary control. The initial meeting was held in 1996 by committed Mum, Kerri Carboon, whose first child was born with PKU. That first meeting in Victoria attracted 100 people representing 14 different disorders. Since 1997 membership has grown across Australia and around the world representing 16 disorders. Most are inborn errors of protein metabolism, though we do welcome individuals and families with other disorders such as inborn errors of carbohydrate and fat metabolism.

Founder and current Executive Director, Kerri Carboon, deliberately named the organization to bring together families not linked by a specific medical diagnosis, but by the main issue for those with metabolic disorders - dietary control. The aim of the MDDA has always been to bring together individuals and families and to work with professionals, organizations, government departments and other agencies to provide a comprehensive resource and information centre so people can make informed choices about their care. The MDDA also has an extraordinary track record in successfully working co-operatively with the Australian Government to ensure a range of services are provided to families and that policies are in place to protect member's interests.

The MDDA became a registered charity and Associations Incorporation in July 2000. Kerri (and long-suffering husband Neill) were successful in obtaining financial assistance from the Australian Society for Inborn Errors of Metabolism (Metabolic Dr's body), Nutricia (lo pro suppliers) and Micromass (manufacturers of MS/MS) to attend the PKU and Allied Disorders International conference in America in May 2001. The MDDA held it's inaugural National Conference in July 2001, and were able to fly parents across Australia to attend this important conference thanks to funding from the ASIEM, Nutricia, Sharpe Laboratories and the Department of Family and Community Services (a Federal Government Department). FACS also supplied funds to have the conference video taped and sent to clinics around Australia. Speakers who may be known to those in America, included Dr. Steve Kahler and Dr. Susan Waisbren. Kerri and Neill also presented their report from the American conference.

In 2000, the MDDA successfully lobbied the Federal Health Minister to obtain a grant for each and every individual with an Inborn Error of Protein Metabolism to assist with the financial costs of lo pro food. This money (\$200 per month) is paid directly to the affected individual or their carer (that's "caretaker" in American English). In 2002, the MDDA also successfully lobbied the Office of Disability to include new criteria in assessment forms for a disability "Carer Allowance". This has meant many parents of children with inborn errors of protein metabolism are now eligible for about \$1500 per year (paid direct to the parent). This payment recognizes the additional financial, social, and emotional needs that metabolic families face when caring for their children under sixteen years of age.

The best news arrived at the end of 2001, when the MDDA received ongoing National Secretariat funding from the Federal Government to continue it's operations and expand it's membership Australia-wide. This really was an endorsement of our good track record in working with governments to achieve the best outcome for those we represent.

I think it's important to acknowledge, how much of this work is achieved by pure persistence in beating your head against various brick walls. The committee all try to slot in

their volunteer work for the MDDA around the rest of their "life" and for many of us, that has meant sacrificing work, family and other important priorities. It has also meant many, many late nights, endless administrative tasks and even a few tears of frustration! Kerri has been known, on a few occasions, to simply keep working throughout the night, and to get up from her computer at 5am to start organizing her kids/day/ordinary life. The rest of us, unaffected by mania, do similarly stupid things - although not quite as drastic!

The mad flurry of activity since receiving the National Secretariat funding is simply too much to note, however, you can now check out our activities at our brand new website: <http://www.mdda-australia.org>. The site is still a work in progress and it's final completion is not expected until January 1<sup>st</sup> 2003. The site has an email listserv, for those of you who need to expand your communication internationally just to find a person with the same disorder as you!

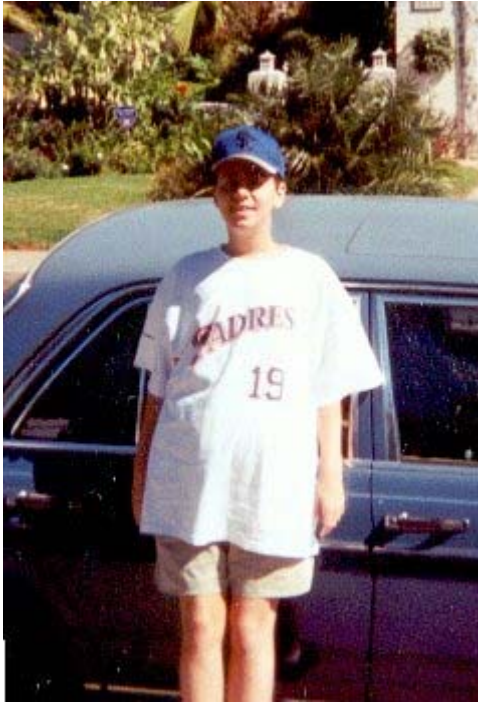
Just like you, we are actively pursuing the important issue of ensuring all Australian babies are properly screened at birth (MS/MS). We have also negotiated with the President of the ASIEM to hold a parent conference in conjunction with the International Congress on Inborn Errors of Metabolism in Brisbane, Queensland, Australia, September 2003. We'd certainly love to see a few international families there!

Be assured that "Rachael, Mum to Sarah, 3-MCC" is continuing personal membership of the OAA (which is just as well since I stole so many fantastic ideas for the MDDA website!). We hope, as two organizations, to enjoy a positive working relationship for many, many years to come!

# Peter Savvas Remembrance

*Propionic Acidemia (7/17/82 – 1/27/02)*

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As told by his father, Minas:

He was the most lovable creature I have encountered. Yes, he was "special needs" (as the current euphemism would have it), but, o God, if we all could be "special needs." He was goodness incarnate, and I never loved anything more than I loved him. His mother loved him as well, of course, and she nurtured him with vigilance and devotion, but I doubt that she loved him more than I did. I was his father and, if he needed it, his mother, his friend and his admirer. Some 300 people (many who had met him for only 10 minutes) came to his funeral. On his tombstone, I call him a "courage-teacher" and I thank him for teaching us unconditional love, patience and devotion.

# Ann-Marie Lynn Jones Remembrance

*Propionic Acidemia, September 29, 1997 – March 11, 2002*

*by Ashley Ham*

The hate and sadness  
Running through your mind  
You looking for some one  
You will not find  
Her body's there  
But she is not  
You know that she's gone  
You miss her a lot.  
Her little hands  
Her body so small  
You're looking around  
Waiting for her call  
Her precious soul  
Was taken away  
Although there's no way  
That she could've stayed  
It may seem like time  
Is going too slow  
Remember the good things  
Where did they go?  
Through your fingers  
She has fallen  
If you listen to the wind  
You'll here her calling  
She'll always be there  
Inside your heart  
She is not with you,  
But you never did part  
She'll always be there  
Where ever you'll be  
For inside you  
Is every memory.  
Forget the bad times  
Remember the fun  
And always remember  
Her life is not done.  
I will always remember the cute way you'd laugh, and the sweet touch of your hand.  
Rest now darling.  
Sleep and dream.  
For now you shall never feel pain again.



We all love you Annie Bananie!

Thank you to the following are individuals who sent donations to the Organic Acidemia Association in memory of Ann Marie Jones: Tina DiBiagio & Family, Rita MacDonald, Marlene Morgan, Tech-Med Incorporated, Pam & Steve Thorp, Area Agency for Developmental Services, The Shea Family, Lynne Wolfe

# A Mother's Birthday Wish

*by Vicki Vanderveen*

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It was twelve years ago today that I became a mother. A tiny, wrinkly, pink baby, with a big voice, made me such. I remember thinking that God had just plucked the wings off an angel, and gave him to me, so perfect this child was.

Looking back, I realize he was observant from his first moments. We locked gazes in the delivery room, and because my arms were strapped down from the surgery, he reached his tiny fingers out and touched MY cheek. He knew we belonged together.

In all honesty, I made the most of Christian James' three years. Not a day went by that I wasn't with him. And I can account for every single second of his life and know that he was, at all times, never more than a few feet away from someone who loved him. Few of us can say we lived our lives like that.

It was such an idyllic life. I was married to my high school sweetheart. We were happy. And two years after we were blessed with our son, a daughter with a Gerber-face and an infectious smile joined the family. It was like a storybook. In retrospect, whatever problems we thought we had, were minor.

I KNEW I was blessed. Still ... I took much for granted.

Then, one day it all changed.

I am grateful — gratitude, as I learned, comes in many forms — that he did not suffer long. It was a brief illness, no longer than a weekend. He had the flu. The hospital saw him, and said that. His doctor saw him, and said that. Most of our friends did not even know he was ill.

Monday, the day after Easter, my husband Larry went to work, as he was improving. Then, hours later, he was dead. Larry would hear that while talking on a borrowed cell-phone, in rush hour traffic, in New York City, while driving. There is no tactful way to tell a man his son has died.

But the first words out of his mouth were, "Oh Vicki! Are YOU all right?" In my darkest moments, I remember that testament of love. Ask me why I stay with this man — plain and simply, those words.

Yes, that suddenly, that chaotically, that simply — he was gone.

As we would find out four years later, it was not JUST the flu, but a complicating metabolic disorder that was undiagnosed — methylmalonic acidemia. Essentially, I watched my son starve to death in front of my eyes, in a matter of hours, and as articulate as I am, I could not find the words to tell the doctors that.

A third of my life I have been a mother now. But I take nothing for granted anymore. Nothing. Not one second. Another daughter, Liesel Karinna, was born with the same metabolic disorder as her brother. Because we are not ignorant to hers, she is not at the grave risk that he was. Still, I know how quickly one of my precious loved ones can be gone forever.

I am strong most days. Those who know me best will tell you that. But I give into the pain this time of year. I will not sugar-coat it, it can be debilitating.

But even so, I still know I am blessed beyond words. You see, as I look at it, I am the most fortunate woman in the world. If that sweet angel was only going to be here on Earth for three years and twenty days, at least he was with me. That is three years and twenty days more than any other woman was ever blessed with.

Today, I will be selfish. I will indulge myself in whatever I feel like. I will give in to tears, no matter when they come. I will look for the little miracles I am open to, and the "signs" my son still sends me.

Where is Larry in all this? We grieve together, and we grieve independently. He understands my vulnerability this time of year. But the same things that bother me, do not necessarily bother him, and vice versa. Yesterday, we spent as a family — no playdates, no work, just the six of us, in memory of the One Who Died. We ate out, we shopped, we bowled — nothing monumental, just enjoyed being together. Me, my husband, and our four surviving daughters, who understand matters of life and death and grief too young.

I have not been myself these days. The anticipation of his birthday is almost as bad as the day itself. Next week is Easter, another bad day. But on that day, I will force a smile, and stuff baskets for my daughters, and dine with the entire extended family — both sides. I do that for my girls, for they still need the magic of childhood. But today, I give in to it.

One day a year, that is all I allow myself. Today is it.

Today my boy is 12.

One more hug. That's all I would like. What I would give for just one more hug — to smell his hair, to touch his face.

Do me a favor, friends. Today, appreciate someone. Whomever it may be. Hug them. Tell them how important they are. Look deeply into their eyes, and memorize every facet of their being that you can drink in. One day, they may not be there, and you will be grateful you did. Gratitude comes in many different forms.

Happy Birthday, Sunshine.

# Michael John Dalton

## *Methylmalonic Acidemia and Homosystinuria, Age 14*

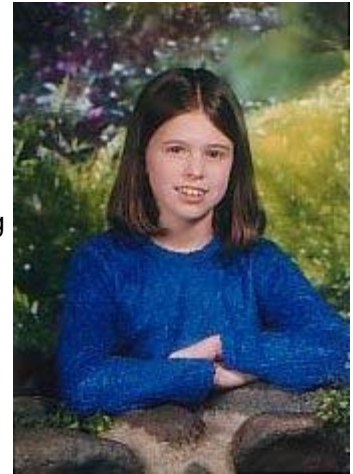
*by Ashley Marie Dalton*

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When my brother was born they did not know that Michael had a disorder called Methylmalonic Acidemia and Homosystinuria. Today he still has the disorder. He is only 14 years old. He cannot talk, all he can say is Ma Ma, Baby and Ba. Sometimes when my Mom calls him he comes. Michael has to have 4 shots a week. I think my brother can understand what we say, like... when my mom says go put your clothes in the laundry basket... sometimes he throws them on the floor on the way to the laundry room. But I am not sure if he can understand us. My brother used to pull my hair, it hurt. Sometimes I got mad and he started crying. He doesn't pull my hair anymore. I wish my brother could talk and play games with me. Like tag or ghost in the graveyard. I wish he could

tease me like a lot of big brothers do. My brother has brown hair and blue eyes. Michael likes to play with his big yellow ball and bounce on it with me. He also likes to run after me when I run away from him. I am 9 years old and in the 3rd grade. I wish I could have a brother or sister that could talk and play with me. He screams so loud, because he doesn't get what he wants, I have to plug my ears.



# Eilidh Ruth Duncan

## *Propionic Acidemia, Age 18 mos*



My name is Ruth Milne and I am the very lucky mother to have Eilidh Ruth Duncan as my daughter, here is her story.

Eilidh was born on November 6, 2000 in Aberdeen, Scotland. I had a perfect pregnancy, my first at forty! The 3.5 kgs (7lb. 12oz.) beautiful baby girl breast fed immediately; "a natural," the midwife said. However, over the next day or so she fed less and less and the midwife said

she was just a lazy eater. She was very quiet compared to some other babies in the ward, but otherwise everything seemed okay.

The nightmare began when she was two days old. Alan commented on her rapid shallow breathing. After a few quick checks, a doctor rushed her away to neonatal unit and said he would contact us back in about an hour. He explained it could be an infection or dehydration. We were upset, but we waited and waited. Little did we know what was to come.

After almost two hours the doctor came back and again explained possibilities, including a metabolic condition. They performed numerous tests, and prepared us for seeing Eilidh, now in an ICU incubator and wired up to lots of machines. A few hours later the Senior Consultant had the unenviable task of breaking the news to us. I knew before he even spoke that the news was very bad. He said they strongly suspected Propionic Acidemia but a urine sample being urgently couriered to Edinburgh and a future skin biopsy would confirm this. I calmly asked him outright if she was going to die and his reply was "You have a very, very sick little girl." We were devastated. I felt so guilty as we had not yet named our daughter. We decided our beautiful precious angel would be Eilidh (Scots Gaelic pronounced Aylay). She deserved a strong Scottish 'Braveheart' name, Alan thought, to help her in her fight.

At initial crisis her ammonia levels rose to 350. We spent two weeks in ICU, with Eilidh winning a place in everyone's heart. A skin biopsy was performed at a week old, later showing she only had 0.2 enzyme activity and was non-biotin responsive. To add to our problems a drug addict Mum stole lots of my belongings including the camera with all the first photos of Eilidh. We were on automatic pilot during this time, learning to do the normal duties of new parents plus all the 'nursing' duties. Eilidh was fed by NG tube a combination of a mineral mix and expressed breastmilk as her natural protein intake.

The following days, weeks and months seemed nonstop sickness due to very bad reflux. Her bolus feeds which would last 1½-2 hrs. each time. At three months old Eilidh became very unwell. Everyone focused on trying to achieve normal ammonia levels. Eilidh's skin broke down as if scalded and was very puffy. She was very anemic and a bone marrow biopsy showed that the marrow had stopped producing red cells. The possibility of a rare blood condition and regular transfusions were discussed. Coincidentally at that time, by increasing her natural protein levels Eilidh 'cured' herself.

Since then, Eilidh has gone from strength to strength. Last summer she decided not to eat for about 7 weeks but we persevered and she switched back on. I remember being delighted when she would eat 5 spoons worth and now this Toot polishes off a couple of jars

at a time! We still mainly use organic pureed baby food, she will eat small amounts of finger foods and now drinks well from a cup.

Last September Eilidh had her first metabolic crisis since birth. It took everyone by surprise as to how quickly things could change. Within 24 hrs. of taking this playful smiling baby into hospital due to sickness and constipation, she was on full IV's and we were being told the maximum treatment was being administered. They reminded us of what may happen and to prepare ourselves. The ammonia levels reached 400 this time. We spent 6 days in the hospital and to everyone's relief there was no damage. It was like a wake up call to not get too confident about the future.

Eilidh is not walking yet, but rolling and beginning to shuffle on her bottom - the physio believes she is capable of much more than she does, and that her very content, placid nature, being very flexible, and Mummy who runs to her every need are the main reasons for delay. We are working on progressing with eating more solids and movement. Eilidh only says a few words but understands everything and I'm sure it won't be long before she is chattering away. Although Eilidh's condition was not picked up by NBS, the hospital's quick diagnosis has saved Eilidh from having any initial crisis damage. We are so thankful to them.

The hospital team is headed by Consultant Pediatrician, Dr. Ian Auchterlonie and Senior Dietitian Kathleen Ross, who are outstanding and excellent in their care and support of us all. We also have 24-hr. open access to the Royal Aberdeen Children's Hospital and all staff are aware of the protocol should Dr. Auchterlonie be out of town (a talent that Eilidh has perfected!).

Eilidh's daily medication currently consists of Sodium Benzoate 3 ml x 4, Carnitine 5 ml x 2, Cisapride up to 2.4 ml x 3 when needed, Soluble Zinc 5 ml x 1, multi vitamin drops 0.6 ml x 1, Lactulose 7.5 ml x 2 and Senna 5 ml x 1. Each month for 10 days she is given 5.7 ml of Metronidazole per day. Diet consists of natural protein in the form of 'normal' food 1gm/kg. XMTVI Analog 650 mls+/day, we are about to change to XMTVI Maxamaid for the night pump feed, plus lots of fluids during the day for hydration. We are currently waiting to have a G-tube fitted, although when well Eilidh takes all daytime food and drink orally.

Everyone is very cautious with regards to infection and only recently have I taken Eilidh into town shopping. I have always maintained that if I can keep Eilidh well enough for long enough then someday a cure may be found. Hence, I have become closely involved in the PAF (Propionic Acidemia Foundation). I realise there are no guarantees with any research, but I am grateful that at least someone is trying to help. I feel I owe it to Eilidh to help her in any way that I can.

Eilidh is the most happy, contented soul anyone has ever met. She gives us so much pleasure and happiness. She smiles and laughs constantly, picking your heart up instantly, should you feel down. She is so full of fun and love.

When leaving the hospital I thought we may only have Eilidh for a few months but she has done so well and surprised everyone. 'Mummy's Little Mouse' we love you more than words can say.

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TV AND DVD TODAY PAGE 19

# Pain behind little girl's smile

FULL STORY: PAGE 9



**TINY FIGHTER:** Eilidh Duncan, who is battling a potentially deadly disease, with mum Ruth. PICTURE BY AMANDA GORDON

... of a child porn probe. Grampian Police confirmed a warrant was executed at a property in the Bridge of Don area. The move was part of Operation Ore which targeted computer users allegedly accessing pay-per-view websites based in the United States. The sites were selling horrific images of sexual abuse of children. Turn to Page 2.

## NE dad to serve prince

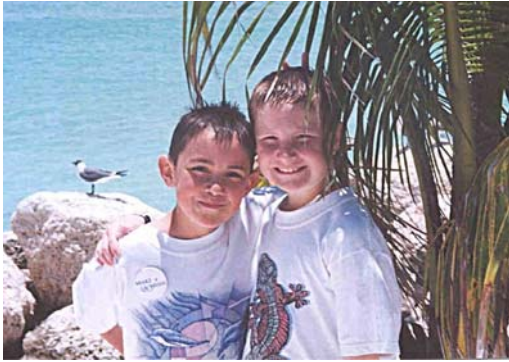
A NORTH-EAST naval officer has been appointed one of the Prince of Wales's key aides. Lieutenant Commander Alistair Graham, 33, will take up the post of equerry on June 5. The dad-of-two grew up near Aberdeen and gained an engineering degree from the city's Robert Gordon University.

Ruth Milne and daughter, Eilidh, PA are featured in their local Scotland newspaper.

# Jonathan Royal

## *Ketone Utilization Disorder, Age 10*

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In the last newsletter, we told the story of Andrew Royal and his diagnosis of Ketone Utilization Disorder in late 2001, at age 11. Andrew's brother, Jonathan (10) was then tested in February, 2002 and was found to also have KUD. After starting the low protein, frequent complex carb diet and Bicitra medication, Jonathan felt somewhat better, but still was quite fatigued and had daily muscle aches/pains. In May, Jonathan was started on Carnitor, and within 72 hours, his fatigue totally resolved. Jonathan also said that his only muscle pains he has anymore is when he "throws too many curve balls in Little League!" He's feeling much better and has even weathered an acute viral illness at home, without having to go in for IV fluids. He's having a great spring and a successful Little League season!

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# Oral Antibiotic Use in Propionic and Methymalonic Acidemias

*by Lynne A. Wolfe, MS, PNP, BC*

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Both Propionic Acidemia (PA) and Methylmalonic Acidemia (MMA) were first described in the early 1960's, and both are known to have severe, fatal neonatal presentations, and milder presentations found in older children or even adults. The enzymes for both disorders are in the same metabolic pathway, and therefore many of the symptoms, and much of the treatment for these disorders remains the same even today. The mainstay of treatment for both these disorders includes strict protein restrictions of four Amino acids: Methionine, Threonine, Valine and Isoleucine, as well as aggressive Carnitine supplementation. However, it is now known that the breakdown of Amino acids accounts for only ~ 50% of the toxic products formed in these disorders. About 25% of toxins are formed by the breakdown of odd-chain fatty acids, Olive Oil being the most common. Another 25% or so, comes from the fermentation of fats and carbohydrates by gut bacteria. A very small amount of toxins appear to come from Cholesterol metabolism, but it is not sufficient to warrant any cholesterol restrictions in PA or MMA patients at the present time.

To date there have been about 10 studies addressing how to lower the toxic build up of acids, in both PA and MMA, that are formed in ways separate from the initial breakdown of dietary proteins. The most studied method is the use of oral antibiotics either continuously or monthly, to keep the bacteria levels in the gut low, and therefore, their contribution of toxic acids in these disorders minimal. In most of the studies, protein restriction of 1.5 grams/kg/day, Carnitine, Biotin, and/or Cobalamin supplements were continued and oral antibiotics added. Vancomycin, Erythromycin, Clindamycin, & Neomycin have all been used, with some decrease in the amount of toxic acids produced by gut bacteria. They are also associated with many side-effects, some bacterial resistance, and do not specifically target the bacterial species known to contribute to the fermentation process in the gut. The most effective antibiotic used was Flagyl at 10-20mg/kg/day for 5-7 days each month. Flagyl targets only the anaerobic bacteria known to be involved in the fermentation of fats and carbohydrates, had the fewest side-effects, and provided the largest, most persistent decrease in toxic acid production of all the antibiotics tested. A drop of 40-70% in the production of toxic metabolites was reported in all the studies. Most importantly the effect lasted 3-5 weeks after the oral Flagyl was stopped. Reported improvements in alertness, behavior, increased activity, less vomiting, less constipation, increased appetite and increased oral food intake have all been reported as positive outcomes associated with oral Flagyl use in children with PA and MMA. The longest follow-ups were two years in length. No antibiotic resistance, increased illnesses, problems with growth, or other difficulties were reported.

It seems likely that adding monthly Flagyl to the regimen of children with PA and MMA could substantially decrease their metabolic toxic load by about 25% with improved metabolic control, generally improved health and well-being, and few side-effects.

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# Combined Methylmalonic aciduria and homocysteinuria (cbIC) - An Update

by Dr. Olaf Bodamer, FACMG

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CbIC is due to an unknown defect of intracellular cobalamin metabolism that leads to secondary methylmalonic aciduria and homocystinuria. The clinical spectrum and age-of-onset of *cbIC* is very heterogeneous. Typically, *cbIC* may be suspected in any newborn or infant presenting with vomiting, feeding difficulties, failure to thrive, microcephaly, developmental delay and neurological symptoms. Diagnosis is typically made following organic acid analysis in urine and confirmed by complementation assay in fibroblasts or in newborn screening by tandem mass spectrometry. Despite the favorable biochemical response to hydroxy-cobalamin as usually observed in all patients, developmental delay continues to be present. This may be explained by the toxic effects of methylmalonic acid and/or homocysteine to the developing brain of the fetus, the continuous presence of elevated plasma and tissue levels of these compounds even after treatment has been initiated and the delay in diagnosis in some patients. The diagnostic work-up should be expanded to include asymptomatic siblings of affected individuals with *cbIC* that need to be screened for the presence of methylmalonic aciduria and homocystinuria.

Patients with *cbIC* continue to be at risk for metabolic decompensation during episodes of intercurrent illness similar to patients with other organic acidopathies. The risk of decompensation for late-onset *cbIC* patients is minimal but is more significant the earlier the patient presented, the more severe the symptoms and the poorer the metabolic control were. It is important for all patients that the parents are trained adequately in recognizing early symptoms of decompensation to allow timely initiation of an "Emergency Regimen" preferably at home or when necessary at the hospital.

Evaluation of metabolic control is currently based on the measurement of plasma MMA and homocysteine levels although they poorly reflect tissue levels and the endogenous production of MMA and/or homocysteine. Using these criteria even well controlled children continue to show slow developmental progress, failure to thrive and frequent episodes of metabolic decompensation. *CbIC* is due to a yet unknown defect of intracellular cobalamin metabolism with impaired formation of methyl- and adenosylcobalamin. Methyl- and adenosylcobalamin are essential cofactors for methionine synthetase ( $N^5$ -methyltetrahydrofolate : homocysteine methyltransferase) and methylmalonyl CoA mutase. The secondary impairment of methionine synthetase in *cbIC* leads to reduced formation of methionine and S-adenosylmethionine which is the most important donor of methylgroups for methylation reactions. Impaired methylation in *cbIC* may be the cause of central white matter disease that is frequently observed and that also is seen in patients with methylentetrahydrofolate reductase deficiency.

In addition microcephaly was present in more than 40% of early-onset patients reported by Rosenblatt and co-workers. The mainstay of treatment is the administration of hydroxy-cobalamin as intramuscular injections, typically 1 mg/day. This therapy should be already initiated when there is a strong suspicion towards a diagnosis of *cbIC* even when complementation analysis has not been completed. Folate and betaine supplementation will reduce homocysteine levels through enhanced remethylation while carnitine supplementation will facilitate the excretion of toxic acyl-groups. Moderate dietary protein restriction to reduce the flux of amino acids through the methylmalonyl pathway is initiated in all early-onset cases to further improve metabolic control.

In conclusion metabolic control in early-onset *cbIC* continues to be a challenge despite adequate, aggressive therapy, introduction of emergency regimens and prompt hospital

admission during metabolic crisis. Late-onset patients are at risk for long-term sequelae when diagnosis and therapy are delayed. Further insight into the biochemical and molecular pathophysiology of this disorder is urgently needed to improve therapy and outcome.

Reference list available upon request.

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# Cambrooke Foods

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The Cambrooke Foods family is pleased to announce the addition of many new products including Home Style White Sliced Bread, Cinnamon Raisin Sliced Bread, Focaccia bread sticks (Italian herb or onion and dill), and Tuscan pizza crusts. MixQuick, a delicious just add water mix for pancakes, waffles, tempura, biscuits and more and an All Purpose Baking Mix are available too. (See our web site for recipe ideas.)

Macaroni and Cheese, Shake 'N' Cheeze, a shaker style premium cheese blend, and Cream Cheese - plain, wild berry and garlic herb have been added to our line of delicious cheese products. Those of you with a sweet tooth will love our new smooth and delicious chocolates.

We recognize your desire to purchase products in smaller quantities and now feature several variety packs.

Try Cameron's Variety Pack, which features favorites like Artisan Bread, Plain Bagels, American cheese, French Toast Energy Bars and Orzo (a tender oval-shaped pasta, great for mixing with vegetables or serving plain with butter). The bagel sample pack offers all your favorites and is a perfect "go with" for the new cream cheese sample pack.

Help a friend in need of low protein foods or celebrate an occasion with a gift of a Cambrooke Foods Gift Certificate — good for any delicious Cambrooke Foods purchase. Check out our website regularly for new products and join our mailing list (easily done by e-mailing us or calling us) so you will be advised of special offers.

Cambrooke Foods has been busy over the last few months not only working on bringing more delicious low-protein foods to your table, but improving customer service, shipping policies and payment options.

Our new Customer Service Manager and our friendly staff are working to make the ordering process easier. You can order 24/7 through our call center, toll-free at (866) 4 LOW PRO, or over our website at [www.cambrookefoods.com](http://www.cambrookefoods.com). If this is not convenient for you, you can mail or fax your orders to us.

# Children's Hopes & Dreams Foundation Pen Pal Program

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Wouldn't it be nice to have your children write to other children to share the common ups and downs of being a child with a life threatening or chronic illness or disability?

The Children's Hopes & Dreams Foundation, Inc. is a registered non-profit foundation which began in 1983. The PEN-PAL Program is for children diagnosed with a chronic or life threatening illness or disability from the ages of 5 through 17.

We have researched and found a great need for a PEN-PAL Program which can give children an opportunity to create the friendships, support, understanding and fun they deserve. Your child's special PEN-PAL will be able to relate to your child in a way no one else can! Their friendship will be priceless! Our PEN-PALS learn from one another as well. Each child can relate their experiences in a different light. Your children will learn how to cope with their own condition better from sharing with another child going through the difficult trials of an illness or disability themselves. The Organic Acidemia Association is now participating in this program – for an enrollment card, please call/email/write to:

**Kathy Stagni**  
**13210 35th Avenue North**  
**Plymouth, MN 55441**  
**763-559-1797**  
**Email:[OaaNews@aol.com](mailto:OaaNews@aol.com)**

# God's Golden Children

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An OAA parent, Katie Martin recently published a book on dealing with the disorder called Crigler-Najjar Syndrome. Katie has two children with this disorder. Captured within this book are 30 touching stories written by parents, children, doctors and scientists from all over the world and how they cope with a very rare, frustrating, genetic disorder called Crigler-Najjar Syndrome (jaundice).

Read about having to sleep under very bright lights for 10-12 hours every night to escape brain damage; being called nasty names because you look a little different; sleeping at a hospital for five years; or thirteen years of loneliness, isolation and fear of the unknown.

The book sells for \$9.95 and you can place your order for one by contacting Katie at:

**RD 1 Box 177-A  
Swengel Road  
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570-966-3814**

# I met a golden angel

*by Maureen Pandorf*

**Sister to Debbie Ottinger and Mom to Charlene, 9, Glutaric Acidemia, Type 1**

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I met a golden angel, she walked upon the earth  
And always glowed with happiness since the second of her birth.

She showed me things I'd never known through eyes so bright and blue  
She taught me how to crawl, then walk in everything I do.

She never uttered any words that would make sense to most  
Yet talks a blue-streak through her smile, but never would she boast.

When life is hard or times are sad and seems like a losing race  
I can look at her and feel the love and she slows my fastest pace.

I met a golden angel, so bright and blonde and fair  
I love her now and always will.  
She's my angel, "Charlie Bear"