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Organic Acidemia Association



OAA Newsletter Family Stories • Family Matching • Family Conferences • Research Funds • National Advocate for Newborn Screening

SPRING 2013 Parent Support, Education & Awareness

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In this issue:

Orphan Drug Act 30th Anniversary

by: *Jana Monaco 2/26/13*

We all entered the world of rare diseases when a child or loved one of ours was diagnosed with an inborn error of metabolism. It is a diagnosis that swarmed us with fear and trepidation, not to mention many questions and an altered lifestyle. The diagnosis of a rare disorder can be daunting and isolating as it takes us down a path of hospitalizations, genetic specialists, dietary adjustments, therapies, etc. However, in today's world, such a diagnosis is far different than receiving a similar diagnosis 30 years ago. With the click of a mouse, we are suddenly linked with many others living with the same disorder, exchanging dietary regiments, therapy ideas, establishing bonds and friendships with one another and even with the medical specialists managing the care of our affected family member. Thirty years ago, little discussion and medical attention was focused on rare disorders; let alone inborn errors of metabolism. Without such focus and internet, people were isolated. People did feel like 1 in a 100,000 as they struggled to understand their rare disorder, find the proper medical professionals that understood their disorder and how to treat it and live with it. The future of the Rare Disease Community changed dramatically when a group of vocal patient advocates spearheaded a move to bring rare disorders to the forefront in the nation's eyes. They worked with government partners, media and others to publicize the unmet needs of those living with rare disorders. Like the large marches that occur in Washington DC today, countless numbers of people swarmed the US Capitol 30 years ago that lead to the signing of the Orphan Drug Act into law by President Ronald Reagan on January 4, 1983. This legislation opened the doors for research and the development of treatments for millions of people in the United States with rare diseases or disorders. Abbey Meyers, the leader in this effort, together with other patient advocates, formed the National Organization for Rare Disorders (NORD), whose mission was to provide advocate, educate and provide research and patient/family services nationwide to those affected with rare disorders.

On January 4, 2013, the FDA Office of Orphan Products Development hosted the first of a series of events to observe the anniversary of the Orphan Drug Act. FDA Commissioner Margaret Hamburg, MD spoke at the event, which included comments from NORD President Peter Saltonstall and Abbey Meyers. As part of the celebration, the FDA selected 30 "rare disease heroes" to honor for their advocacy efforts for the rare disease community.

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Medical Advisors

EDITORIAL

As we start a new year, I have heard from so many of our families got hit hard with illness and hospitalizations. We were not immune to this as Melissa got hit with that nasty rotavirus that was going around. Fortunately just a couple of visits to the ER was all she needed. Hope everyone is staying healthy and a hoping for an early Spring!

This issue if the newsletter is packed full of great articles that range from our newest families to our veteran warrior children. I am also sad to share a couple memorials from a couple of brave OAs that have joined our group of angels in heaven. It just never gets any easier to hear of a death of one of our OA "family." I am honored to share all of these stories in the newsletter.

We also are so excited to have quite a few new clinical studies for our organic acidemias! Never have I have heard of so many researchers looking for participants for their research studies! So exciting! I encourage you to consider bringing your child(ren) to any of these studies -- it will help further the research into our organic acidemias.

And last -- but not least we have decided on the location for our next FOD/OAA family conference --- thanks to the support of Children's National Medical center we will be in Washington DC. Stay tuned for details.

Peace, Kathy

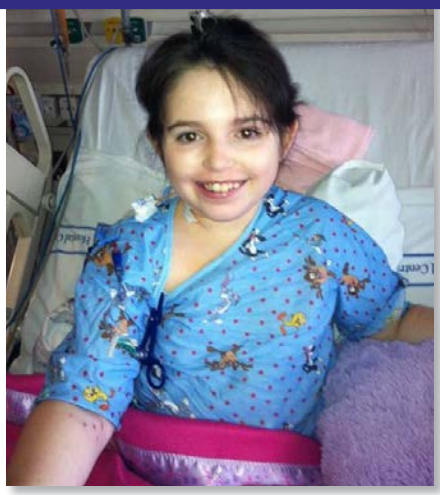
YEARS 30

Orphan Drug Act continued...

This list of heroes included patient advocates, legislators, researches, clinicians and industries. Our own, Jana Monaco, Advocacy Liaison for the Organic Acidemia Association was one of the patient advocates recognized as a "rare disease hero" for using her experience with her children Stephen and Caroline and their diagnosis with isovaleric acidemia, to advocate for rare disease issues such as newborn screening, medical foods insurance coverage and neurodevelopmental disabilities awareness. The Rare Disease Community has earned this monumental opportunity to celebrate the many accomplishments and life saving products achieved and developed over the past 30 years.

by: *Jana Monaco 2/26/13*

Kaitlin : Propionic Acidemia | 13 years | Liver Transplant Recipient



Kaitlin Three days post transplant

Our daughter Kaitlin was born in 1999 with Propionic Acidemia. For 13 years she had numerous hospitalizations, fed 100% through a g-tube and used a wheel chair if we needed to be out and about. We had a few really good "healthy" years where Kaitlin did really well and was getting stronger and things were looking up. In 2010 she started heading downhill yet again. We couldn't keep her ammonia levels down and she became increasingly lethargic. Her lethargy became so bad that she had to use her wheel chair almost all of the time. Then in 2011 we approached our metabolic team about what we could do for Kaitlin as her quality of life was declining. This wasn't the lively little girl that we knew and it was heart breaking. We discussed medication and treatment options and one of those options was a liver transplant. My husband and I had a big decision on our hands. We set up an appointment to meet with the transplant team at Seattle Children's Hospital. The initial consultation was incredible. The transplant doctor was so at ease discussing Propionic Acidemia and his knowledge of the disorder was incredibly reassuring. As most of us know finding anyone in the medical field who had knowledge of metabolic disorders and can discuss it without glancing at a sheet of paper with facts about the disorder is nearly impossible! This man spoke to us like he knew what it was like to have a child with PA. We were "sold." We went back a few weeks later for the full workup for the transplant. It consisted of lots of labs, ultrasound, and interviews with the docs, social work and the list goes on. At the end of the second day of tests the coordinator said she would call us and let us know if we were approved by the transplant team for Kaitlin to have her transplant. It was a nerve wracking wait and I can't remember now how many days it took but the day we got the phone call that she was a candidate was a happy and scary day all wrapped into one. We signed all the necessary paperwork to have her put on the list and we got the call she was listed for a liver in October of 2011. That year we celebrated Halloween, Thanksgiving, Christmas and her 13th Birthday waiting for the BIG call. On January 11th 2012 we got the call to bring her in that a liver was on its way for her. Of course my husband was out of town, I was in an employee review meeting and both my kids were in school….So I did what any good mom does…Have a full blown panic attack! Between hyperventilating and crying I had to have my employee call my business partner to come pick me up so that I could get Kaitlin from school and to the hospital. My husband was now on his way home from his trip and the whirl wind of pre-transplant began. Labs, ultra sounds, and lots of waiting to hear if the donor liver is viable. Early the next morning they came into the room and told us it was time. The liver was a perfect size and match and they were ready to get started. I will tell you that Kaitlin has had lots of surgeries and hospitalization but this was by far the hardest time letting her go back with that team leaving her in their hands.

Fast forward 12 hours...Surgery went incredibly well with no complications. She was in ICU starting to wake up and we were about to experience having a new child all over again. She was taken off formula that day and was given just Pedia Sure. All her metabolic meds were stopped over the next week and the LOADS of transplant meds were started. After transplant the amount of medications is overwhelming! A few months prior to transplant Kaitlin had to start a thyroid medication and we taught her to take pill by mouth. I am glad that I did that! After her surgery she had so many pills to take it she did it like a champ. There was one medication that is an oral liquid that has to be swished in the mouth and swallowed four times a day to help prevent yeast infections. This was the hardest med to get her to take! She did however learn to do it and after a month would just suck it down.

Kaitlin had a couple rounds of rejection which isn't uncommon after transplant. Since March of 2012 all of Kaitlin's liver labs and metabolic labs have been normal. Going into transplant her ammonia was three times normal and post-transplant it was in the single digits! We had never seen that in her 13 years. For the last year Kaitlin has been formula free as well as g-tube free. She eats 100% of her food by mouth now. Some of her favorites are noodles, rice, shrimp, steak, and chocolate cupcakes.

For the last year we have gotten to know a whole new kid. Before transplant she was always kind of dazed and in a fog. Now she is witty and funny and talks all the time. She can tell us about her day in great detail which is so exciting. She has learned to ride a bike and loves to "race" us running. Her endurance is what has been the biggest change. Before transplant she could barely walk from the couch to her room without being tired. We had to push her in her chair if we left the house. In December of 2012 after transplant Kaitlin was granted her Wish from Make a Wish and we went to Disney World. She walked everywhere, stood in lines and then ran when she would see a princess. Just seeing her be able to have the endurance and stamina of a typical kid is one of the largest blessings we could have asked for.

[next page...]

[Kaitlin...]

I get the question all the time if it "cured" her propionic academia…She will always have PA but no longer shows any symptoms of the disorder. We traded one set of issues for another. We still have to be very diligent about medications, fluid intake, calorie intake and labs. But the stress of PA is no longer there for us. We feel as if a large weight was lifted from our family and the quality of life for all of us has changed tremendously. We live life to the fullest and there isn't a day that goes by that we do not send a prayer to the family who had to have a significant loss for us to have a gain. We are eternally grate full to the donor family, the surgeons, the medical staff, family and friends who have supported and guided us through the last year.

by: *Michelle, Trent, Kaitlin and Maddy – Seattle, Washington* | jacqulinemichelle@hotmail.com

Chesia Isovaleric Acidemia 10 months

Italy – translated from Italian

My name is Ciro and with my wife Damaris would like to tell you the story of our little child Chesia. She was born on March 2 2012 at the Hospital Guglielmo da Saliceto of Piacenza (Italy) with a natural birth. Soon she was dismissed from the Hospital and we waited same days for the result of the screening she was submitted. After some days we noticed that the baby had problem to be breast fed and that she slept a lot night and day. We called the gynecologist, but she said that we should not worry. On March 7 the doctor Giacomo Biasucci called, and told us that the baby was positive to the neonatal screening for Aciduria Isovaleric we were worried for the life of our daughter, but we called upon the name of our master and savior Jesus Christ and realized his peace in our hearts. We were not informed about this pathology so we made a research on the internet, but we found very little information about it. In Italy there are three regions only that perform neonatal screening, and we thank God that in the region we live this type of exam was introduced on March 2011. Doctor Biasucci is still struggling to introduce this type of exam in all the Italian Hospital.

Chesia started sleeping more and more so the doctor decided to administer L' Carnitine 1 ml 3 times a day. From the blood test the doctor noticed the hyperammonemia of 222, 0/ mg but, before the evening the value of the ammonia was normal. Thanks to the timely diagnosis and an appropriate therapy Chesia has reported no physical consequence and no secondary biochemical alteration. In the following days there were no longer signs of drowsiness, and Chesia began feeding with breast milk. The baby has continued to take L-Carnitine 1g per Kg every day with the addition of glicina 200 mg per kg every day and to food mixtures (Iva Anamix And Duocal). Back home was like reliving the birth of Chesia, although with many questions in our minds. Chesia was fine and her smile gave us the strength to take care of her.

Chesia now is one year old and she grows regularly. She eats vegetables with pasta and rice (protein -). Each goal she achieves we can see the hand of God. We actually know that there is no cure for this type of pathology, but we hope that God can give wisdom to the researchers to find solution or makes a miracle. In any case our daughter will have a not normal life but a special life. In a few years we will have to explain to Chesia her pathology and make her understand that she is not a sick girl but only different from the others. To be different doesn't mean to be sick.

I take this opportunity to thank: Prof. Giacomo Biasucci, Doctor Giuseppe Gregori and Doctor Angela Pozzoli

We would like to create with your help a web site about organic metabolic acidemia. We hope to see on your web the picture of our little Chesia with others children. Finally we want to thank your organization for all the news that you send to us. May God bless you.

by: *Ciro ,Damaris ,Chesia and the little dog Pluto.*
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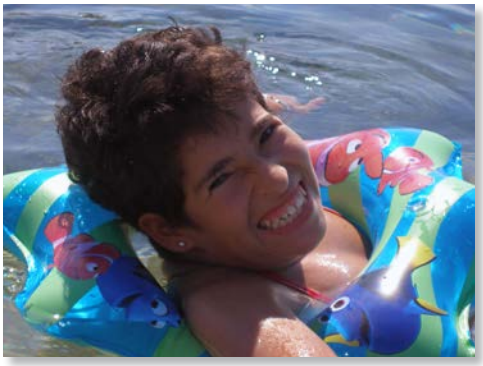
Memorials



Claudia Pintus – MMA, Mut 0
October 16, 1990 - March 15, 2011

Our little angel, Claudia, MMA mut 0 has been called at the home of Father at the age of 20 y, 5m. She was an adopted baby, abandoned at birth in hospital. She was a happy person with a real passion for life. She had a lot of friends. The chorus of her school has been entitled to her name.

Stefano e Angela Pintus
Cagliari, Sardegna, Italy

Matthew – Propionic Acidemia
Jan 16, 1985-Dec 16, 2012

For years we have been receiving your newsletter, and each time reading the stories of families.

We lost our youngest son, Christopher, to complications from PA almost twenty years ago. The day we buried Christopher we discovered that our oldest, Matthew, also had PA. Through the years he lived with courage, grace and a smile.

His PA lead to cardiomyopathy, which eventually lead to heart failure. While suffering the insult of PA, and heart failure and being on the heart transplant list, he managed to complete his degree in Industrial Engineering at Dalhousie University in Halifax, Nova Scotia and got his engineer's Iron Ring.

This summer his appendix burst which lead to catastrophic results. He had a Left Ventricular Assist Device implanted and slowly clawed his way through 3 months in hospital. He was waiting for a new heart. Sadly one never came; he died from a cerebral hemorrhage on Dec. 16, 2012.

All these years of receiving the newsletter, I always was afraid to write of Matthew, thinking that we were doing ok, that I could not share his successes with the other parents, because we were not going to lose. Hubris.

Matthew had always wanted to go on an expedition with his Cardiologist, Dr. Heather Ross of the Peter Munk Cardiac Centre at Toronto General Hospital.

Though he could not go with Dr. Ross on her South Pole 2013, she did take his Iron Ring with her. Dr. Ross' inspiration for this trek was Matthew. Read about her emotional blog post here: *http://inaheartbeat.ca/tyl/?p=734*

This is a picture of the marker at the South Pole. Notice Matthew's engineer's Iron Ring on the top of the sphere. The ring was carried to the South Pole by his doctor,

Dr. Heather Ross of the Peter Munk Cardiac Centre, of the Toronto General Hospital. Dr. Ross and the *www.testyoutlimits.ca* team made the trip to the South Pole to bring awareness to the need for additional research in heart failure and heart transplant, as well as the need to *www.beadonor.ca* to register to be an organ donor not only in Canada but throughout the world. Had a heart been available last September, October, November or December, Matthew may have survived to still be with us today.

Order Your 2013 OAA Calendar

and other related items and help the Organic Acidemia Association

http://www.cafepress.com/organicacidemia

Thank you again Raymonde DeGrace who once again did an outstanding job designing our 2013 Calendar cover....and thanks to the many families who sent their photos to include in this project.

NIH Mini Study

DO PEOPLE WITH ORGANIC ACIDEMIAS HAVE NORMAL IMMUNE FUNCTION?

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VIRAL ILLNESSES AND IEM.

Sometimes it starts with a fever, a sniffle, or a cough. Other symptoms such as nausea, vomiting and diarrhea may also occur. Different types of viral illnesses occur throughout the year whether during the cold winter months (e.g. influenza) or the hot days of summer (enteroviruses). Although many patients may weather these illnesses well, certain high-risk groups, such as patients with inborn errors of metabolism (IEM), are at greater risk for increased morbidity and mortality. Certain patients with IEM seem to take longer to clear infections, may become infected with unusual organisms, or may take a while to bounce back from their illnesses. Some of this may be attributed to their metabolic dysfunction, however, it should be noted that many IEM have been described as having some level of immunodeficiency.

THE IMMUNE SYSTEM AND INBORN ERRORS OF METABOLISM (IEM).

IEM, especially organic acidemias (OAs), have been described as displaying deficiencies in immune function. For example, in OAs (e.g. methylmalonic aciduria, propionic acidemia, and isovaleric acidemia) reduced numbers of white blood cells and antibodies can be found. Since white blood cells and antibodies help the body fight off infection, patients with reductions in these critical components of the immune system may be at risk for serious infections. There are two likely mechanisms by which immune dysfunction may occur in IEM. First, enzymes deficient in IEM may also be deficient in immune cells. This may lead to a block in metabolism that is critical for immune system function. Second, toxic metabolites may build up and have damaging effects on immune system function. Toxic metabolites such as lactic acid and ammonia are known to inhibit the function of immune cells. While there are at least 13 IEM that have been identified as having some form of immunodeficiency, the scope and depth of the problem is under-characterized.

NUTRITION AND THE IMMUNE SYSTEM.

The management of IEM oftentimes involves restriction of offending dietary components, such as protein, which may lead to nutritional deficiencies. Patients may display biochemical (e.g. decreased prealbumin) and physical signs of malnutrition (e.g. hair loss, poor growth). Nutrient and vitamin deficiencies may also coexist. A recent review of patients with phenylketonuria, an amino acid disorder, described suboptimal nutritional outcomes following treatment.1 Besides growth impairment, deficiencies in vitamins (B6 and B12), micronutrients (iron, zinc), essential fatty acids, and protein intake (decreased bone mass and density) were described. These nutritional deficiencies may affect immune system function.

Deficiencies in energy status, protein, vitamins and nutrients, alone or in combination, can lead to clinically significant immunodeficiencies. Nutritional deficiencies at critical periods of maturation of the immune system may hamper its development leading to an immunodeficiency.2,3 In addition to immune system development, proper nutrition is also critical for maintenance of the immune system. For example, deficiencies in various nutrients including protein, zinc, iron, vitamin A, arginine, citrulline and glutamine may affect white blood cell function. The adequacy of nutrition and immune function is highlighted in elderly populations. Elderly patients often have numerous nutritional deficiencies, which may affect their ability to produce protective antibodies after the flu shot. This reduced vaccine efficacy may be overcome by nutritional supplementation and optimization.4-6 These and numerous other studies suggest that proper nutrition is essential for immune system function.

THE NIH MINI STUDY: METABOLISM, INFECTION AND IMMUNITY.

Since infections can trigger life-threatening acute metabolic crises in children and adults with IEM, especially in those with OAs, we have decided to characterize the function of the immune system in patients with IEM. The standard of care for IEM patients is routine vaccination for childhood and seasonal illnesses. However, there have been no studies to investigate whether the response to childhood and seasonal vaccination is normal in OA patients. Vaccination represents a challenge to the immune system and can tell us how well it may be functioning. IEM patients may have enzyme deficiencies in their immune cells, a build-up of toxic metabolites and nutritional deficiencies, all of which may impact immune system function.

The NIH MINI Study: Metabolism Infection and Immunity in Inborn Errors of Metabolism (www.genome.gov/mini) is an exciting new study at the NIH Clinical Center (clinicalcenter.nih.gov). The main goal of our study is to learn about the function of the immune system in metabolic disorders, especially OAs.

There are two ways to participate:

1. Travel to the NIH Clinical Center in Bethesda, Maryland for an evaluation. Travel costs will be provided for patients and their families. Additional visits may be suggested dependent upon the level of subject participation. The visits will typically be 2-3 days long. At the first visit, we will perform a physical exam and do a detailed nutritional and immunologic assessment for all study participants. We will offer Hepatitis A vaccination, which is part of the current Pediatric Vaccination Schedule (*http://www2.aap.org/immunization/IZSchedule.html*). These recommendations are relatively recent, coming in 2005, and many children over the age of 8 years may have missed getting vaccinated against Hepatitis A. Therefore, you or your child would potentially benefit from receiving this vaccine. During "flu" season, the influenza vaccine is recommended for all children with chronic illness, including those with OAs, and will also be offered. As part of the assessment, we will measure whether or not you or your child's immune system was able to respond appropriately to vaccine(s). This includes vaccines that are offered as part of the study as well as past vaccines received during childhood.

Additional tests may include:

- body composition testing, including a DEXA scan
- energy expenditure testing

2. Donate blood and tissue samples to the study. For individuals who are unable to travel to the NIH Clinical Center, blood and tissue samples may be donated to the study. Tissues of interest include: fibroblasts from skin biopsies, and materials acquired from other medical procedures including lymph nodes, tonsils, spleen, and especially cord blood. The acquisition of these samples will be coordinated with your local medical provider(s).

The NIH MINI team is available to discuss eligibility for this protocol with anyone that may be interested in participating and welcomes all inquiries.

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Changing nature of Vitamin D deficiency 2012

by: Lynne A. Wolfe, MS, CRNP, BC

INTRODUCTION

Recently Vitamin D has gotten a lot of interest because of its function beyond bone health. With the recognition that Rickets could be cured with vitamin D its role in the prevention of disorders associated with bone health has dominated the research to date. However, new research has identified vitamin D metabolism may be different in different cell types, is involved in calcium and phosphorus homeostasis in multiple tissues and important in other body functions. It is now known that vitamin D levels positively correlate with conditions such as cancer, immunity disorders, diabetes, muscle disorders and cardiovascular disease.

The vitamin D metabolite 25-hydroxy vitamin D (25-OH) is one vitamin D status indicator. The serum level is determined by skin synthesis through sun exposure and/or dietary intake. Sun synthesis of vitamin D has been limited by changes in sun exposure because of concerns about skin cancer. Skin synthesis of 25-OH vitamin D is limited by skin pigmentation, age, angle of the sun, poor air quality, and the percent of the skin exposed to direct sunlight at any given time. Wearing long sleeves or staying in the shade reduces vitamin D synthesis in the skin however the use of sunscreen, even with high sun protection factor (SPF), did not block the skin's ability to synthesize vitamin D.

Today, dietary contributions of this vitamin are most important and foods, such as Orange juice are now fortified with Vitamin D. Vitamin D3 is considered the metabolically active form of vitamin D and supplements should be with vitamin D3 not Vitamin D2. The scope of vitamin D deficiency is a new focus. Two publications have identified the problem specific to children and adolescents within the US. On the basis of a sample of US children aged 1 to 11 years; millions of children may have suboptimal levels of 25-OH vitamin D, especially non-Hispanic black and Mexican American children (Pediatrics 2009;124:1404–1410). A disproportionate burden of vitamin D deficiency in the non-Hispanic black adolescent population. Females and overweight adolescents are at increased risk (Pediatrics 2009;123:797–803).

ENDOCRINE FUNCTION

For the endocrine functions of vitamin D, the kidney is the main site for changing 25-OH vitamin D to its active metabolite 1,25-OH vitamin D. Once made in the kidney, this active metabolite enters the blood stream and is circulated to all cells and organs in the body. The two main functions of circulating 1,25-OH vitamin D are; 1) to increase intestinal calcium and phosphorus absorption, and 2) to stimulate bone metabolism. Other important functions include; decreasing renin production in the kidney that helps to regulate blood pressure and increasing insulin secretion in pancreas that helps keep blood sugar levels normal. Other organs (muscles, colon, prostate, immune system and pancreas) can also change 25-OH vitamin D to 1,25-OH vitamin D.

IMMUNE SUPPORT

Vitamin D is important for stimulation of normal immunity. Current research demonstrates that 1,25-OH vitamin D improves the function of several white blood cell types that fight infections. It also turns on several pathways that make proteins that can destroy bacterial cell membranes.

CANCER PREVENTION

1,25-OH vitamin D appears to stop the growth and differentiation of cancer cells. It can also support apoptosis – cell death in cancer cells.

MUSCLE HEALTH

Vitamin D deficiency has been associated with muscle atrophy (wasting) and poor muscle contraction. 1,25-OH vitamin D helps muscle cells maintain normal calcium balance. Calcium is essential for normal muscle contraction and relaxation. 1,25-OH vitamin D also improves muscle cell growth and function by interacting with multiple other hormones and enzymes.

Vitamin D has a benefit on muscle function in hyperparathyroidism that can cause several muscle tissue and functional abnormalities.

HEART HEALTH

Vitamin D may be beneficial for preventing cardiovascular disease. Hyperparathyroidism is associated with high blood pressure, 1,25-OH vitamin D can indirectly impact on high blood pressure by decreasing parathyroid hormone levels. Vitamin D also interferes with the renin–angiotensin system (RAS) that regulates blood pressure by decreasing renin production. Some evidence also suggests that vitamin D status can impact on heart muscle contractions as well.

WHAT LEVELS OF VITAMIN D ARE NEEDED?

Two types of vitamin D are currently described "deficiency" and "insufficiency". Deficiency corresponds to a level of 25-OH vitamin D level below 25 nmol/L, a level that

was set to prevent rickets or osteomalacia. The Institute of Medicine (IOM) defines the desired serum 25-OH vitamin D level at 50 nmol/L for promoting bone health. This level is considered too low to support the functions of vitamin D beyond calcium metabolism and bone health.

Vitamin D insufficiency is suggested by increased serum parathyroid hormone and markers of bone turnover, decreased intestinal calcium absorption and osteopenia by DEXA scans. Effects related to vitamin D insufficiency can be masked serum 25-OH vitamin D levels between 25 and 75 nmol/L. Thus, serum levels of 25-OH vitamin D in this range can be used to define a state of subclinical vitamin D deficiency. More than 70% of individuals can be at risk of subclinical vitamin D deficiency. Levels of 25-OH vitamin D above 75 nmol/L may be necessary to maximize musculoskeletal benefits. Several lines of evidence suggest that levels of 25-OH vitamin D above 75 nmol/L are associated with beneficial outcomes without toxic effects, such as elevated serum or urine calcium levels.

The US and Canadian governments requested the Institute of Medicine (IOM) to update its 1997 report on Dietary Reference Intakes (DRIs) of calcium and vitamin D so in 2010, new Recommended Dietary Allowances (RDAs) were published and recommended between 600 and 800 IU/day as "values sufficient to meet the needs of virtually all healthy persons". There considerable debate concerning what daily intake should be as the importance of vitamin D beyond bone health is getting more attention. Some researchers suggest 25-OH vitamin D levels should be above 75–80 nmol/L to support immune function and cancer prevention.

The US Endocrine Society recommends an intake of at least 1,000 IU to raise the blood level of 25-OH vitamin D consistently above 75 nmol/L.

It is also generally recommended that high-risk populations that include obese children and adults, as well as children and adults on anticonvulsant medications, glucocorticoids, antifungal medications like ketoconazole, and many medications for AIDS should increase their Vitamin D3 intake by at least two to three times the recommendation for their age group.

Current scientific evidence suggests that 25-OH vitamin D serum levels should be over 75 nmol/L; otherwise, there is no beneficial effect beyond bone health. Though there is an ongoing controversy about changing the recommendations for vitamin D supplementation at this time. It seems apparent that the current RDA's are based on preventing Ricketts and maintaining bone status in primarily healthy children and adults; not for maintaining general health in children or adults with chronic illnesses who may also be on medications, such as seizure medications, that can have a negative impact on Vitamin D metabolism.

References are available upon request.

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Maggie Grace : Isovaleric Acidemia | 7 months

On June 27th 2012, Maggie Grace was born, the only little girl on both sides of our family. With her unexpected red hair and bright blue eyes, she was the most beautiful girl I had ever seen.

The day after she was born was perfect. Friends and family came to the hospital and cuddled her for hours without her making a fuss. Not even her super-excited three-year-old brother could coerce a cry from her. At the time, we thought she was simply a very easy-going baby. My only nagging concern was that she wouldn't nurse. Experts marched in and out of the room during every feeding with limited success, but told me to keep practicing and that she would soon catch on.

Day three was when everything changed. Since Maggie had been so mellow the day before, my husband Chris left the hospital to run some errands. The nurse came in to take Maggie so I could shower and eat. She commented on how Maggie felt a little cold, but did not express any concern. The morning came and went. I finally called the desk to remind them that it was time to feed the baby, and they told me she had been taken to the NICU. I dropped the phone and started down the hallway as fast as I could go. My cell phone was in my pocket, and I called Chris to tell him that when he finished with what he was doing he needed to come on back to the hospital. What he heard was hysterical crying and gulping with no intelligible words at all.

[next page...]

[Maggie Grace...]

At first they thought she might have some kind of infection. A close friend who happens to be a pediatrician called me often so she could help interpret the tests they were doing. It sounded routine. I kept thinking that anytime Maggie would start to improve, and we could take her home. But she wasn't responding to the treatment. Her temperature was too low; she wouldn't eat and wouldn't stay awake. We watched our cherub-like angel shrivel into a little grey sickly looking baby, and there wasn't anything anyone could do about it. Based on some of the testing, the neonatal pediatrician began to suspect that Maggie was suffering from some sort of metabolic disorder and recommended we move her to Children's Medical Center. She had been so kind and caring with us, I could hardly believe she was the same woman I heard barking to someone on the phone about rushing Maggie's newborn screening results.

The next couple days were a blur as we waited on the newborn screening results. Endless experts and specialists flowed through the hospital room offering opinions and explanations. Wonderful nurses offered comfort. I lost more than 20 lbs. while I watched Maggie waste away. We couldn't hold her because of the IV in her belly button, but we stayed with her in the room when allowed and slept in the visitors' area when the doctors were with her. When the newborn screening results came back, the attending physician met with us and explained the diagnosis. He told us Maggie had a rare genetic disease called Isovaleric Acidemia and explained how the disease prevented her from processing leucine protein. The disease is very manageable and, once treatment started, Maggie began to improve immediately. When she cried, the nurse tried to shush her, but it was music to my ears since she previously had been too weak to cry.

She came home July 11 and, as of today, she continues to thrive. Her disease is managed by a strict diet that allows a limited amount of leucine protein. She shows absolutely none of the negative side effects many children suffer when they go into metabolic crisis.

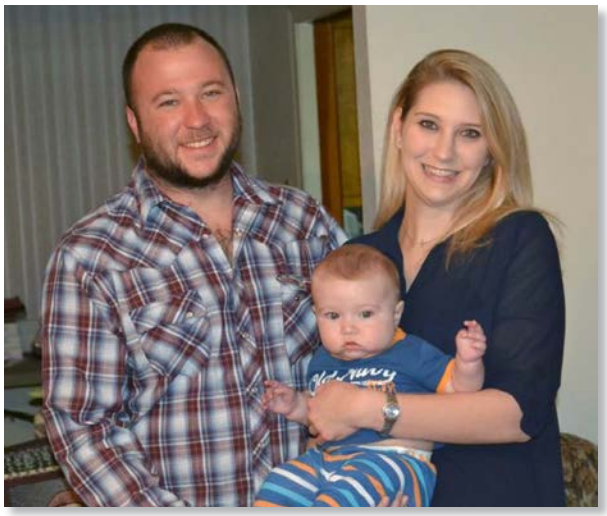
Maggie is almost 7 months old now and growing like crazy. At her six-month, well-baby visit, she was in the 80th percentile for height and weight. She is a very happy baby and loves interacting with people.

While most babies would have started solid food already, IVA babies start solids a little later. We go in for lab work every week so they can monitor her leucine levels. She has had plain rice cereal a couple of times, but we have to wait to hear what our doctor has to say about the latest lab results before she can try anything else. The biggest challenge so far has been educating our family! Maggie is a seventh generation Texas princess. We Texans like our BBQ so our family is having a hard time understanding that Maggie can't have things like that.

Despite having to do things a bit differently from other babies, Maggie is in excellent health and that can be credited 100% to the newborn screening that allowed for early treatment. We are eternally grateful for every day we have with our baby girl.

by: T*rish – Parker, Texas* trishcook1@yahoo.com

Bradley : MMA Cbl C | 6 months



Bradley was born August 2, 2012. He was a week over due and was born a healthy 7lbs 12ozs and 20 ½ inches long. My husband and I were so happy to finally hold our beautiful baby boy. It took us over a year and a half of trying to conceive and one miscarriage before we were blessed with Bradley in our lives. I had the best pregnancy and delivery ever. I had absolutely no morning sickness or any major complications. I did have one overnight stay in the hospital at 25 weeks due to placental lakes, but it did not cause any harm to me or Bradley.

Bradley was examined by his pediatrician, Dr. Sanches, shortly after birth and scored a 10 on his APGAR assessment and passed his hearing test as well. I felt so relieved to know that my beautiful baby boy was completely healthy. I started breastfeeding right away and he did great despite him being tongue-tied. We were sent home after the normal 48 hour stay in the hospital and was told to do a follow up in a few days with our pediatrician due to Bradley having a little bit of jaundice. At the follow up, our pediatrician was concerned that Bradley had not reached his birth weight yet. I was told to supplement with formula and continue to breastfeed and come back in a few days. We continued to do the follow ups every two days and Bradley just wasn't gaining weight. I eventually gave up on breastfeeding and went to 100% formula, but he still wasn't gaining weight.

In the middle of Bradley not gaining weight, my husband and I were really concerned with his irregular breathing and the way he choked every time he ate. We were told the irregular breathing was completely normal in newborns, but recommended we see a radiologist and a speech

therapist to conduct a swallow test to make sure he wasn't aspirating the formula. The swallow test showed that he was not aspirating, but just hadn't learn to swallow properly yet and that him being tongue-tied had nothing to do with his swallowing. They recommended we see a surgeon for him being tongue tied because it would become a speech problem later on in life and to start putting rice cereal in every bottle because the thicker the formula the better it went down. At first I followed their recommendations on the rice cereal, but after only 2 days and no changes in the choking I stopped and realized something else was going on, but just didn't know what. During all the daily doctor visits, we were also going back and forth to the lab to redo Bradley's newborn screening test because the first test came back abnormal. We were told that it is normal for the results to come back abnormal and that usually the second test will show that everything is normal. Well, the second test came back abnormal as well, so we were told to do a more extensive testing. And when those results came in I will never forget that phone call because it forever changed our lives.

Bradley was exactly 3 weeks old and it was a Thursday evening around 5:30. My husband and I were playing with Bradley and the phone rang, it was Dr. Sanches, our pediatrician. I just knew something was wrong because it was after office hours and the actual doctor is calling me not his nurse and I could hear in his voice something just wasn't right. He said the results came in and it showed that Bradley had Methylmalonic Acidemia, MMA. He said we needed to get to the Children's Hospital in New Orleans immediately where the genetics team is waiting on us. My heart just sank and I started crying hysterically and the whole time my husband is staring at me knowing something is wrong, but I just couldn't get it out to tell him.

When we arrived to Children's they immediately sent us back to do blood work to confirm that he did in fact have MMA and to start treatment immediately. Being that the hospital is over two hours away from our home we did arrive there late and the Geneticist, Dr. Michael Marble, was already gone for the day to discuss the disorder in detail. In the meantime, lots of teams of doctors came in and out to evaluate Bradley throughout the night and they were all shocked to see him so healthy. By the next morning, Bradley was already eating four ounces of formula instead of two and he had stopped choking when he ate as well. It was so crazy how fast the treatment was working. That morning we met with Dr. Michael Marble and he discussed MMA in detail and went over the treatment with us. Bradley's treatment consists of Hydroxcobalamin IM injections, Levocarnitine, Folic Acid and Betaine. We also met with Dr. Marie Dalme, the dietician, and she went over Bradley's diet. He drinks Propimex-1 twice a day along with his regular formula. We were sent home that day due to how great Bradley was responding to treatment.

Today, Bradley is almost six months old and very happy and healthy. He is growing so much and all his doctors are pleased with how well he responds to treatment. Since diagnosis we found out the sub type of MMA was CblC. Bradley is in therapy as a preventative measure and as of right now he seems to be right on track as far as development wise. We are so blessed to have Bradley in our lives. He is definitely our little Hero!

by: *Jena* *JGRodriguez@hotmail.com*

Chad : Propionic Acidemia 20 years

Wow, ten years have passed since I last wrote an update on my son, Chad, PA. My last article was in the June 2003 issue.

As I try to figure out where to begin, I guess I will start by a brief introduction. My name is Sandy, single mom to two boys, Michael, age 27, and Chad, 20 years young in May 2013. Only Chad has PA as the boys have different fathers. We live in Hilo, Hawaii. Both boys were born and raised here. Hilo is a small town on the Big Island. Over the years I have thought of moving some place that would possibly offer more opportunity for the boys, but we have an awesome support network here and I truly can't imagine living anywhere else.

Michael graduated, with honors, from the University of Hawaii at Hilo with a BA in English. Currently he works with Chad as his home hospital tutor and afternoon personal assistant. This year he is working on applying to Cornell for his graduate studies. I am VERY proud of him. Being raised in a single parent home with a disabled brother has had its challenges and opportunity to see life in ways most aren't blessed to live. Hence, he is a poet with insight on life that he hopes to share with others.

As I mentioned Chad is home hospital tutored. This is the second time in his public schooling that we have utilized home tutoring.

[next page...]

The first was when he was elementary age, at the time he was too fragile to attend school and I guess I was fearful of what could occur in the public setting. At that time he had an indwelling Hickman catheter as I was running IV fluids and TPN on him daily. The fear of infection was always on my mind. Currently, he is home tutored due to falling at school. His first fall was March of 2012. This last fall was September of 2012. More on these falls later.

Looking back, the years from Chad's tenth birthday to around age 15 were great. No significant illnesses, only occasional colds that we managed at home and generally lasted about a week or so. Potty training happened during this time, BIG relief, and we were more active than ever imagined. Nearly every day, weather permitting we went to the beach. Chad loved going so much that even days when it was a bit chilly Chad still demanded we go to the water, (Chad has a sound he uses to communicate water). On those chilly days he still got in the ocean while I stood there freezing!! No Chad doesn't swim, he uses a "swim ring", and as his confidence grew he asked to go out to the waves and bubbles. That is out quite far where folks are body boarding and surfing, I took him but always prayed for safety. Not knowing how long his life will be, I continually try to find and offer him new adventures that he can manage. We also spent many weekends sailing with my friend on his Hobie cat, (a 16 foot catamaran sailboat). Chad loves sailing, raising his arms to feel the wind, dragging his hand in the water, even getting sprayed as we "fly" through the waves, he giggles with delight.

Then during his 15th year, he began signs of puberty. It seemed as if overnight he started getting taller and growing hair in places he never had. With puberty and his rapid growing, his bones became compromised. With his low muscle tone he just doesn't have enough mass to protect his bones. Therefore, a series of "breaks" began. First, he had slipped in his bedroom; a hearty sneeze knocked him over, resulting in a hairline fracture in his humerus. Not wanting to cast it, due to possible muscle atrophy and protein breakdown, we kept it stabilized for six weeks, (still went to the beach). Then, a few months after that healed, I noticed him beginning to limp. After a couple sets of x-rays, orthopedic exams, and finally a bone scan, we found a stress fracture in his femur, just above his knee. Again not wanting to cast him, we dealt with leg braces for about 7 weeks. Also he was not to put weight on it, ok, so now back in a stroller, he is gaining weight, about 72 pounds now and standing 4' 6", and I am barely 5' and maybe 98 pounds. This was VERY challenging. By God's grace we got through that only to take yet another fall about 6 months after his leg healed. This time it was his knee. Back in a leg brace, this time for about 5-6 weeks. This last "break happened in March of 2012. So during all these bone incidents he is continuing to grow taller and gain weight, finally. Just about two weeks into this last period of being in a brace while at school his aide went around the corner to get his stroller and he tried getting up out of his seat, with a brace on, and fell on his face. Nothing broke, but his chin and cheek were bruised pretty good. His brother was picking him up from school that day as I was working. Well Michael, (brother), immediately drove him to where I was working, Michael arrived, ten minute drive, and I still had no call from school. Michael was seeing "red", angry beyond belief. Finally school called, no one saw what happened!!!! So, the next day we took Chad to the doctor and she recommend he not go back to school until his leg was out of the brace as, "they obviously can't handle him". Once he was finally out of the brace, before I sent him back to school I called a meeting with the entire team in his classroom. I outlined EVERYTHING about Chad and propionic acidemia, as it seemed they didn't "get it". With the school year just about over, Chad returned for about a week.

This brings my update to June 8, 2012. This morning started as any other, Chad woke about 7am, called "mommy", I responded, "good morning little man, mommy will bring you medicine". I drew up his Allegra, walked to his room with the syringe, and he out of the blue went into his first gran maul seizure. In panic I ran to get the phone, he was safe in bed, his pediatrician didn't answer, seizure still happening, so I called 911. The ambulance arrived quickly and we spent the ENTIRE day in the ER. July 3rd was his second seizure, no ER this time, but that is when we started him on Keppra. He has had a few seizures since we started the med, but the last couple of months he has been seizure free. One of the mild seizures was while in the ocean so he is now reluctant to go in the water, but still loves kayaking and hanging at the beach watching waves and the big splashes!!

Backing up a bit, school started August 26. He was to have a new aide this year, he receives 1:1 services, this aide has been in the classroom with him for the last two years so I feel ok with this gentleman working with him. After all he was at my little safety meeting just five/six months ago. Well on September 5 as I was preparing to pick Chad up from school and take him to water therapy, the phone rings, "Sandy, come to school right now, Chad has fallen in the bathroom". I respond, "Is he ok?" the answer back is please come right away. I flew out the door and arrived in five minutes; it usually takes me seven or eight minutes. In the classroom bathroom, there is Chad sobbing with a mouth full of blood and his top lip swollen. I remained as composed as I could, for Chad. After looking him over and comforting him I

realized he just wanted out of there. We left and Chad has not returned to the class and NEVER will. That evening after his bath I noticed both knees were severely bruised, and one was very swollen. That was the last straw for me. I contacted an attorney and thus far there is no lawsuit, but believe me I got everything I wanted at the IEP meeting requesting he be tutored at home. We are still watching and waiting to see if his teeth will die and need surgery or if they will live and he will just live with chipped teeth for his lifetime.

As for his medical status, still growing, now about 4' 10", 82lbs, and beginning to have facial hair, (not into shaving him, that's just one more thing to do). He is still receiving all his nutrition via g-tube every three hours and overnight. His formula consists of prophree, polycose, pregestimil, propimex-2, cyclinex-2, and his meds are: sodium benzoate, buphenyl, poly citra-k, Carnitor, neomycin, biotin, Keppra, Allegra, lactulose, and of course a vitamin, Thera-plus. Chad has his own language that is easy to learn once you know him, he loves his companion/therapy/service dog, a black lab, and having his brother as a tutor is pretty awesome. His favorite outing is going to our quaint airport where he walks back and forth checking the elevators and watching people hide behind doors, he could care less about the airplanes. It is only at the beach that he enjoys watching the planes fly off into the distance. Our lives are quite simple and complicated at the same time, always revolving around Chad and his wants and needs. Chad has a new weekend aide that he loves, a good friend of mine for over thirty years, she has been a godsend.

Life is good, exhausting at times and I give God all the glory and praise for orchestrating our every day. We are on FB, just recently, and I am trying to learn how to post "stuff", so that I can share with others. If you are ever in Hawaii, give us a call: 808-959-3030. Aloha and blessings to all our OAA friends and families.

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Organic Acidemias and Energy

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As many of you know, organic acidemias (OAAs) are life-long disorders which affect many organ systems. My research focuses on trying to understand the effect of the propionate pathway disorders, predominately Propionic acidemia (PA) and also Methylmalonic acidemia (MMA) on energy production, especially the Kreb cycle (also known as tricarboxylic acid (TCA) cycle, or the citric acid cycle). However, the other OAAs often have similar energy problems. Patients with these disorders can have myopathies, metabolic stroke-like episodes, cardiomyopathy, and eye problems. These are symptoms of energy deficiency problems.

In vitro studies in my lab are trying to determine if cell lines from individuals with propionic aciduria have differences in the amount of protein that makes up certain enzymes in the Kreb cycle compared to cell lines from people with more typical biochemistry. We think this is the case. We are also looking at how well those enzymes work in these cell lines. In addition, we are also examining whether external stress on these cell lines change how well these enzymes work and how much of these enzymes are present.

In addition, we are starting a study that looks at resting metabolic rate (RMR, the amount of energy one needs when at rest) in individuals with OAAs (and related disorders) when they are sick and when they are healthy to determine whether RMR changes with illness in OA and whether this is similar to findings in individuals without these disorders. Presently (due to the importance of measurements in individuals when they are sick), we are doing this at our own institution, Children's National Medical Center in Washington DC. If results are promising, we will try to expand to other clinics. Preliminary data are promising at this point. This is a way to look at whole body energy production in people as opposed to just cell lines.

Finally, with the goal to gather information about individuals with OAs, Children's National Medical Center is also a site for the European registry and network for Intoxication type Metabolic Disorders (EIMD). EIMD is gathering information about individuals with Methylmalonic aciduria (MMA), Propionic aciduria (PA), Isovaleric aciduria (IVA) and Glutaric aciduria type I (GA1). The website for the organization is http://www.e-imd.org/en/index.phtml. If you are interested in participating and your metabolic clinic is not presently involved, I would be happy to send you and your metabolic doctor information about this study. It is predominately a chart review. Send me an email with your contact information and your metabolic doctor's information (KChapman@childrensnational.org), if you are interested.

Thanks to you all for your support and time.

Lucas : MMA Cbl C | 3 years

Lucas is a three year old boy. He has amazing blue eyes that twinkle when he is being mischievous (they twinkle frequently) and shaggy blonde hair! He likes to play trains, be read to, watch Curious George, wrestle with his brother, and boss his sister. Lucas rides his balance bike at break neck speed and has mastered all playground equipment. He has a great sense of humor. He can be completely stubborn and incredibly sweet (sometimes at the same time). Sometimes he is shy; sometimes he is a wild child. He also has Cobalamin c disorder (Cbl-c is a rare genetic disorder that can be fatal without treatment). Fortunately he is responding beautifully to treatment but he is slowly losing his fine vision and he cannot talk yet he communicates beautifully. Cbl-c is part of who he is but it does not define him.

Recently his special day class went on a field trip to the pumpkin patch. Lucas was super excited to see all the pumpkins so we were up ahead of all the other kids. A guide was telling a few school employees about photo opportunities. A para (aka teacher's aide) said, "You know they are…." My mommy senses tingled as I knew nothing good could come next. In that short second that she tried to find the right word, I willed her to say kids, preschoolers, wiggly (anything that described all young kids). Of course she didn't. She said nothing. Instead, she finished "You know they are" with a flick of the wrist. To me this said that they are not quite right, a bit off, a tad iffy. In one gesture, she not only marginalized my child, she did it to every child there!

This gesture left me speechless which is not something I am accustomed to. Some would say I am a bit too outspoken but this time words failed me. I am not sure if it was anger or sadness that caused the inability to speak. Tears welled in my eyes. I was stunned by how profoundly this affected me. If you asked me how I was doing being a special needs mom, without hesitation, I would say "Great" and continue on about how fortunate we are to have such an amazing kid. But this wasn't about me. This was about a thoughtless, simple gesture. I know the person who said it did not mean for it to be hurtful. The fact that it was unintentional did not make it any less painful. The fact that she worked with this group of children made it even worse. She should have known better.

I regret not saying something to the person at the time. She does not work with Lucas so I probably will not see her again. I hope by sharing this story people will stop and think before they speak. Most of all, I hope people will realize that a disability is part of what makes a child who they are. The child is not the disability!

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Organic Acidemia Imaging Study

Children's National Medical Center
Washington, DC

Objective: To correlate dysfunctional brain networks in relation to white matter (myelin), biochemical, and cognitive abnormalities in patients with organic acidemia

Problem/significance: Patients with organic acidemia have a history of metabolic brain injury that on the mild end may cause working memory and executive function disorder, whereas more severe cases are associated with autism and severe mental retardation. Dr. Gropman's lab has been using noninvasive MRI studies to unravel biomarkers of neural injury in these disorders which will lead to identification of earliest markers of disease and can be used to develop and monitor new therapies.

Although the exact mechanisms of brain injury are not completely known, previous research has disclosed that pathogenic mechanisms leading to white matter injury that underlie cognitive deficits include cerebral edema, energy failure and neurotransmitter alterations and basal ganglia injury. An understanding the underlying causes that contribute to the cognitive phenotype will allow one to determine which functional networks are affected and whether there is there a link between cognitive, metabolic changes and affected networks.

Eligibility: Patients with Organic acidemias of any type are eligible to participate if they are between 7-60 years of age and can understand instructions in English (need not be native speakers). They must not be ill at the time of study. Females must not be pregnant and must agree to take a pregnancy test. They must not be claustrophobic or have metal devices in the body. The scans are done without sedation so subjects must be able to lie still for at least 15 minutes at a time.

Travel expenses, hotel and a meal stipend are provide as well as pictures of the brain.

If interested email
Organic.acids@gmail.com

SERC-GMDI Nutrition Guidelines Project update

Keiko Ueda, MPH, RD and Elaina Jurecki, MS, RD

The Nutrition Guidelines Project is a multi-year project that was developed out of the acknowledged need for the development of evidence and consensus based nutrition guidelines for medical nutrition therapy (MNT) for people living with rare inborn errors of metabolism (IEM) e.g. propionic acidemia (PROP1), maple syrup urine disease (MSUD), medium chain acyl Co-A dehydrogenase deficiency (MCADD), and phenylketonuria (PKU). The project is funded by a Maternal and Child Health Bureau Health Resources and Services Administration (HRSA) grant administered by the Southeast newborn screening and Genetics Collaborative (SERC) and staffed by experienced dietitians representing the professional society, Genetic Metabolic Dietitians International (GMDI). The project objectives include:

1. Developing a systematic methodology to critically and objectively gather and review both evidence and consensus based information. IEM specific evidenced based information includes; available IEM disorder specific peer-reviewed published research studies as well as available IEM disorder specific gray literature such as; non-peer reviewed IEM medical nutrition therapy (MNT) book chapters, metabolic clinic IEM protocols, and IEM MNT educational lectures. IEM specific consensus based information, gathered from surveys and meetings of clinicians experienced with managing these disorders, is also vital due to the lack of IEM evidence based information2.

2. Developing an internet based portal to direct and document the development and progress of each nutrition guideline. This allows each guideline to be transparent and revisable to all members of the work group.

3. Organizing and recruiting experienced and expert metabolic dietitians and physician volunteers to participate in the development of nutrition management guidelines.

4. Publishing the IEM specific nutrition management guidelines to standardize IEM specific medical nutrition therapy based on current evidence and consensus based information.

Given the complexity and variability of each IEM, along with the paucity of scientific literature regarding treatment and management of these conditions, this project has presented many challenges. But we are working through these challenges due to the importance of developing these guidelines, and we appreciate the patience of our patients and their families who are anticipating the publication of guidelines. We are looking forward to the publication of nutrition management guidelines for Maple Syrup Urine Disease (MSUD) developed with this new evidence-consensus based method on the GMDI website in the near future. Currently efforts are ongoing to validate and update the existing GMDI MCADD and VLCADD nutrition management guidelines, and to develop nutrition management guidelines for PKU and Propionic acidemia.

We will keep you posted on our progress.

1. US National Library of Medicine, Newborn screening coding and terminology guide website: *http://newbornscreeningcodes.nlm.nih.gov/nb/sc/query?reportDefault=reportConditionDetails&conditions=conditions&applications=applications&submit=go* accessed Feb 2013

2. Singh RS, Rohr F, Splett PL. Bridging evidence and consensus methodology for inherited metabolic disorders:creating nutrition guidelines. J of Evaluation in Clinical Practice 2011

Organic Acidemia Association
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Organic Acidemia Association (OAA) provides information and support to parents and professionals dealing with a set of inborn errors of metabolism collectively called organic acidemias. The OAA is a volunteer organization registered with the IRS as a 501(c)(3) non-profit corporation. Donations to the OAA are tax deductible. OAA publishes a newsletter 3 times a year, hosts a Google Group for information exchange and maintains a website. Services are funded by corporation & individual membership donations. Annual membership donation of $25 (US) and $35 (international) plus $5 for the family roster is requested, but not required. Our 501(c)(3) non-profit status qualifies OAA for United Way donations through their write-in option. If there is a write-in option, just write "Organic Acidemia Association" in the blank line on your pledge card. Donations can also be made at OAA's website through the "PayPal" and the "Network for Good" option.

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OAA Internet Google Group

OAA's main mission is to empower families with knowledge about organic acidemias. If you would like to connect with other families who share the same or similar diagnoses, please join our private OAA Group. Visit the *OAANews.org* web site to sign up.