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Organic Acidemia Association



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

FALL 2012 **Parent Support, Education & Awareness**

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It's been a busy year so for OAA – In April, I represented OAA at the GMDI Conference in New Orleans. We shared a table with the FOD Family Support Group. It was a wonderful opportunity to create awareness and share information with dieticians from all over the world.

Our 1st OAA Awareness Month was this past June. Our black & white Charity Spotlight banner ad was in the June 22nd USAToday News section. Another great way to create awareness to our rare disorders. Here's a link to the full ad - http://www.oaanews.org/images/USATodayCharitySpotlightJune2012.jpg. Our newsletter banner (above) is based on color version of our ad:

Well another FOD/OAA Conference is in the books – we had a fabulous conference in Portland in July. Over 200 FOD/OAA families attended from all over the world. I want to especially thank Deb Gould – my partner in planning our conference – this is the 4th conference we have planned and hosted with the FOD (Fatty Oxidation Family Support Group). Deb and I work very well together and very comfortable making sure the logistics are taken care of. Special thanks to our main sponsor, **OHSU Human Genetics Initiative and the Department of Molecular and Medical Genetics** – who assisted us with not only financial support, but also supplied wonderful speakers and volunteers for our child activity room.

OAA Families: July in Portland, OR

I also want to acknowledge and thank OAA mom, Janet Reimers…who assisted me and Deb tremendously – from checking out the hotel prior to signing a contract – to shuttling boxes, bagging t-shirts and shopping for low protein treats. I want to thank another OAA mom, Raymonde DeGrace who did a wonderful job creating our ending ceremony slide show -- all the attendees loved it – Here's a link to the slideshow - http://www.youtube.com/watch?v=MOp5CLVbF3Y. (See photos from conferences throughout this issue)

We really had some outstanding speakers at the conference this year. OAA was able to sponsor eight families to travel to the conference. For those who could not attend, the presentations are posted on OAA's website http://www.oaanews.org/2012ConferencePresentations.htm Our next conference will be the summer of 2014 --- maybe in Washington DC.

by: *Kathy Stagni*

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Financial Statement

As is our custom, the board of the OAA publishes the organization's annual financial status each fall following its filing of the annual 990 statement as required of all non-profit entities. Below is a summary recap from that filing.

For 2011, contributions were down 40 percent and program expenses lower by 50 percent from 2010. This is typical for the organization during "non-conference" years when our spending needs are less. Major expenditures during the year included a one-time metabolic outreach research grant to Children's Hospital of Pittsburgh and a major upgrade to our internet web site. We ended the year with $43,410 in the bank which provides the funding necessary to execute the national family conference that we just held in Portland.

OPENING FUND BALANCE	
OAA Operating Fund	$20,621.21
OAA Research Fund	$13,344.00
TOTAL	$33,965.21

CURRENT YEAR CONTRIBUTIONS AND INTEREST	
OAA Operating Fund	$27,562.66
OAA Research Fund	$4,386.04
TOTAL	$31,948.70

PROGRAM EXPENSES	
Newsletter	$5,824.64
Representation at Conferences	$3,896.11
Accounting Services	$1,650.00
Site Redesign/migration	$5,192.41
Office, Internet, Phone, etc.	$1,405.46
Misc. Expenses	$1,405.46
TOTAL	$19,503.01

RESEARCH GRANT	
Children's Hospital of Pittsburgh – (For Metabolic Outreach project)	$3,000.00

ENDING FUND BALANCE	
OAA Operating Fund	$28,680.86
OAA Research Fund	$14,730.04
TOTAL	$43,410.90

On behalf of everyone associated with the OAA we once again thank all of our financial contributors – without their support, we would not be able to provide the support services for parents and professionals that has become the hallmark of our organization for over a decade.

more FOD/OAA Conference photos


IVA Families: July in Portland, OR

"The 2012 OAA/FOD Conference was such a huge experience for my son Caiden and me. We are thankful we got the opportunity to attend and will be going as much as possible in the future! I learned more from the speakers and parents than from just research and his doctor's appointments. I also found other food companies we can order from and tasty recipes to use. Best of all he got to meet other kids just like him!" IVA parent from Texas


D2HGA Families: July in Portland, OR

OAA and FOD Exhibit at the GMDI Conference (Genetic Metabolic Dietician International) in New Orleans in April

OAA and FOD had the opportunity to share information with metabolic dieticians from around the world at their conference in New Orleans on April 19-21.

Thanks to Menta Pitre, (mom to Ashtyn, MMA Cbl C) for helping out at the booth during the conference.

Yusuf: Methylmalonic Acidemia, Mut 0 | 9 years old

As the flight took off from Kuwait International Airport, my wife and I looked at the adorable little fellow snuggled comfortably in the bassinet. We were returning from a month long trip to India after visiting family with our 9-month old boy, Yusuf. We were glad that we were heading home to US, so we can settle back into our daily routine.

We were totally not prepared for what the future had in store for us. About an hour into the flight, my wife noticed that our little one was displaying a strange breathing pattern. We noticed that he was breathing fast and then he suddenly started to cry uncontrollably. My wife took him into her arms and tried to console him but in vain. As his crying slowed down, there was a remarkable slowdown in his breathing as well. Now he was breathing very shallow. The stewardess made an announcement and luckily a doctor, who was travelling to London, came to our rescue. By now Yusuf's condition had deteriorated rapidly and he had his first (and the only episode) seizure. The doctor immediately conferenced in the crew and explained to them the seriousness of the situation at hand and that the baby needed immediate medical care. The crew was able to get permission to make an emergency landing in Istanbul, Turkey. While all this was going on, my wife and I had lost every hope of saving our son and we were frantically praying with Yusuf unconscious and pale on my wife's lap.

In no time our flight descended at the Istanbul airport and we were on the ground with the emergency crew waiting for us. An ambulance whisked us to the hospital. Yusuf was admitted in the Critical Care unit of the International Hospital, Istanbul. The next hour seemed like the longest hour of our lives as we waited impatiently for a word with the doctors. Finally one of the doctors emerged and updated that Yusuf was very acidic with dangerously low blood sugar and was still unconscious. They would not say whether he would make it or not. And to add to that they would not let us in to see our precious one for full four hours.

Separation anxiety – the height of it - we experienced while waiting outside the CCU. Finally we were let in to see Yusuf. We rushed into the unit and tears flowed down uncontrollably looking at our baby hooked up to all kinds of medical devices and catheters. My wife was surprisingly stronger than me (which I had always known for a fact)... After sitting at his bedside for a few hours, one of the staff members signaled that it was time for us to leave the unit. Half-heartedly we left the unit and headed upstairs to the patient room which was arranged for us by the hospital administration. The next few days were a roller coaster ride; while our son was slowly improving and had regained conscious, the doctors were still not able to get to the diagnosis. All this changed on the sixth day when the doctors took us into the office to break the news. Our son, Yusuf has been diagnosed with a rare form of metabolic disorder called, Methylmalonic Acidemia (MMA). This was based on the lab results that were done by the state of the art labs in Ankara. While the diagnosis was a blessing, the rarity and the non-treatment of the condition left us speechless. It was hard to fathom the future and the prognosis. And I was still in denial.

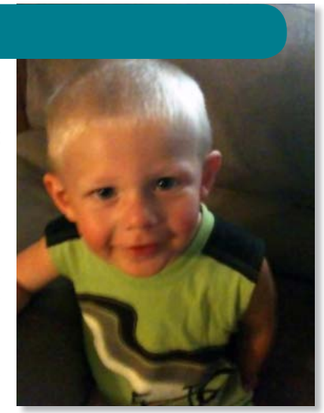
After having spent twelve days in Istanbul, we were finally allowed to travel back to the US with a medical crew. Yusuf had been stabilized but was on a feeding tube. As soon as we landed in the US, we drove straight to the A. I. Dupont Hospital for Children, Wilmington DE. After multiple tests, the diagnosis was confirmed. A central port and G-tube was surgically placed and Yusuf finally made it home exactly one month since he left our ancestral place in India.

Yusuf is now an adorable boy, who will turn 10 in November. The challenges have been and still are many. From monthly lab work to speech therapies, feeding therapies at Kennedy Krieger Institute, patient studies at the National Institute of Health, Yusuf is now ready to celebrate his 10th birthday on November 18th. He is at a regular school but has had some challenges in terms of coping up with other kids. We are also exploring options for his liver and kidney transplant.
Since then we have been blessed with three other children – two girls, Mariam (6 years – unaffected), Khadija (3 years – MMA Mut 0) and and a boy, Ibrahim (1 year – unaffected). Khadija also goes through the same therapies, dietary restrictions and special formula. She is still struggling to gain height, weight and other development milestones.

Living with two special needs children has definitely changed our lives. Despite all the challenges, our precious kids keep our lives in perspective. Frequent hospital trips and hospital stays has made us enjoy and appreciate small things in life and has made us more thankful for everything. We begin our day with lots of hope and carry on with our chores and try to act just like any other parent in the social gatherings. Throughout the day, we are however constantly reminded of the challenges that our kids face. Keeping our face straight under these circumstances is very difficult but nonetheless it has to be done. Because we cannot let Yusuf and Khadija feel that they are any less abled but rather they are differently abled. And yes, I have made that leap from denial to acceptance.

by: *Irfan & Farheen, Newark, Deleware* ipatel72@gmail.com

Dylan James 3-MCC 3 years old



Dylan James was born on June 3, 2009 at 5:59 pm, 1 minute after his twin sister, Mya. He weighed 4 lbs. 7 oz. and was 16 ¾ inches long. I was so ecstatic to hear they were crying and were perfectly healthy, just small. This pregnancy was rough; contractions started at 24 weeks confining me to bed rest for the remainder. The next 3 months I was in and out of the hospital with contractions until preeclampsia at 37 weeks sent me for an emergency C-section. Mya and Dylan spent no time in the NICU and actually had discharge papers before I did.

A week later, I receive a call from the pediatrician's office explaining to me that some of Dylan's newborn screenings came back abnormal and that the pediatrician wanted to see him as soon as possible. We scheduled an appointment and the next morning the pediatrician informed me that my son had an Organic Acidemia disorder and that we needed to do further blood work. I said "An organic what???" I had never heard of such a thing. The amazing thing was that his twin sister, Mya did not have any abnormal newborn screenings.

We did another round of blood work, again same results. The pediatrician advised me to take him to Shands HealthCare in Gainesville to meet with a geneticist. Dylan and I met with his geneticist and genetic dietician. The geneticist gave him the most thorough examination I had ever seen and then went over all of his results with me. He explained the elevations in his urine and blood work were consistent with 3MCC. They were stumped though because his elevations were only 2 to 5 percent higher than normal instead of the 10 to 20 percent elevation they had seen with other 3MCC cases. With this result, we were instructed to do more blood work and urine analysis. Once the blood was drawn, we drove back home and waited for the results.

About two weeks later I received the call that Dylan's blood work and urine analysis was consistent with the other results and that he should begin taking .75 ml of L-Carnitine twice daily. He was not put on any diet restrictions because studies had been showing that limiting the protein intake in some of the other 3MCC patients was actually making those patients protein deficient. Dylan was on Good Start formula and ate the same foods that his older sisters did. He has always been in the small percentile rank among other children his age for his weight and height, falling in the 3 to 5 percent, but the doctors did not seem concerned by this.

In March 2010, the geneticist took a small piece of Dylan's skin to grow cells from it so the enzyme levels could be studied. A couple of months later we received a call from the geneticist, and once again he was stumped. Out of the three enzymes studied, all enzyme activity came back within normal limits. Shands HealthCare called the University of San Diego, where the cells had been sent, and asked them how many times had a child with 3MCC had come back with normal enzyme activity. Their response was that out of 300 cases studied; Dylan was one of three that came back with normal enzyme activity. The genetic team dubbed Dylan their "Medical Mystery" because they have not seen any other case like his where elevations were consistent but on a lower level and enzyme activity is normal. Our next step was to do DNA Sequencing on Dylan's MCCC1 and MCCC2 genes. These are the two genes where mutations can exist, causing the defect and hence giving the term 3MCC. In August 2010, we received the results and found that Dylan has a mutation on his MCCC1 gene. The mutations are G45E and V439L. Dylan is still dubbed their "Medical Mystery" because they have never seen this mutation before. The genetic counselor believes that the mutated gene runs in both families for a very long time and that it finally took Dylan's dad and me to combine them.

Overall Dylan is a healthy three year old. We have been to the emergency room several times, once for pneumonia and all others for unexplained high fevers. The high fevers, the highest being 105.5, always led to a stay in the hospital for a couple of days so he could be monitored while blood work was run. Out of four stays in the hospital, he has only had to be put on dextrose IV once when his sugars dropped below 70. In this instance the geneticist was not too concerned about the drop.

Dylan's high fevers were not explained with every visit in the hospital. All tests came back normal; he was well hydrated, showed no sign of any bacterial infections, and his sugar levels were always normal. We know that Dylan's body can tolerate high fevers and knock on wood, have not had any bouts of diarrhea, vomiting, or refusals to eat. We do not know how Dylan's body will react when he is very ill. I have been instructed to double his dose of Carnitine if he gets sick.

I do not rush Dylan to the doctor or ER every time he has a high fever. I treat him as a normal child until I see something that is not normal with him. That is when I take him to the doctor. Dylan's new pediatrician was surprised that Dylan has no mental delay as far as the other 3MCC patients he has seen or read about do. This is not to say that all children with 3MCC will have some type of mental delay.

Dylan is not on any special diet and eats what everyone in the house eats. The only thing that he seems to have in common with other 3MCC children is that he is on an L-Carnitine supplement. I should note though that Dylan did not have his L-Carnitine for a week and

[next page...]

[Dylan...]

|became lethargic and uninterested in his normal activities and at times stared into space. He began taking his L-Carnitine again and all of those symptoms disappeared. I find it very interesting that although there is no medical proof that L-Carnitine helps children with 3MCC, when Dylan does not take it on a daily basis I see signs that he is not acting his normal self. When the supplement is given to him regularly, he is as normal as any other healthy child is.

After many blood tests, enzyme activity studies, and finally DNA sequencing, the official diagnosis is that Dylan has 3MCC. The doctors believe though that he has a very mild case. He sees his geneticist once a year, at which time they do more blood work and go over any concerns and his development. His geneticist is happy with his development so far and that they do not have any reason for concern. There is still so much to learn about 3MCC. Dylan will grow up with a team of doctors and will help aid in the study of 3MCC especially seeing that his case is very different from other 3MCC cases that exist. Nevertheless, for now, Mr. Dylan is as active and healthy as any other boy his age and he keeps his mom on her toes.

by: *Kristin, Youngstown, FL*
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OAA Families meet and greet in Austin, Texas
August 2012

Connor Methylmalonic Acidemia CblA 10 years

My husband Peter and I live in Long Island, NY. Our first child Connor was born on October 14, 2001. He was 7 pounds, 3 ounces and 20 inches long. He was without a doubt the most beautiful baby I had ever seen. I could not stop staring at this perfect little baby, with his bald head and big blue eyes staring back at me. His Apgar's were 9/9 and he nursed immediately with no trouble latching on. All was good for the first 3 days of his life.

Because Connor was born after 10:30 PM, we were allowed to stay an extra day in the hospital. This extra day was what ended up saving his life! The morning we were being discharged from the hospital and ready to come home, the pediatrician came in, checked Connor and wrote the discharge papers. My husband was on his way to come pick us up to bring us home. I asked the nurse to take Connor back to the nursery so I could finish getting changed and packed up before we left. Just as my husband arrived to take us home, the doctor came into my room and told me that Connor had a fever and trouble breathing, so they were not sending him home. The cleaning lady in the nursery was emptying the garbage and noticed that Connor was struggling to breathe. She immediately got the nurse who called the doctor in.

They initially thought he had meningitis, but the spinal tap was negative. Before we knew it, an ambulance arrived to transfer him to North Shore University Hospital. We were not allowed to go in the ambulance with him, and had to follow in our car. By the time we got there, he was already admitted to the NICU, was on various monitors and had IV's in both arms. The doctors told us his condition was critical and they did not think he would survive the night. They did not know what was wrong with him, only that his labs were off and he had an extremely high ammonia level. As they were discussing Connor's lab values and possible causes, a metabolic doctor who was there seeing another baby who had PKU overheard them and started to review Connor's chart. He immediately began working Connor up for a metabolic disorder. Dr. Alfred Slonim became Connor's doctor and still is almost 11 years later.

The first few days in the NICU were really touch and go. His labs kept plummeting and we were told at least 5 different times that he was not expected to live, and if he did survive he would be severely handicapped due to the very high ammonia levels. Connor had a muscle biopsy done and sent off to Canada. After a few days in the NICU we were told they were going to put him on dialysis. Then his next ammonia level came back and it was slightly lower than the last one. Another repeat level showed an even lower level. They did not put him on dialysis and continued to monitor his labs.

His urine test for organic acids came back at the end of the week and his methylmalonic acid was very high. He was diagnosed with MMA and we were told about the various types. We could not believe what we were hearing! We had to wait 2 days before he could get an injection of hydroxycobalamin because the hospital didn't have it and had to order it in from another hospital. He was given the shot and then we had to wait 3 days to get the results to see if he would respond to the B12. After what seemed like an eternity, the results came in and Connor was B12 responsive. He started getting daily injections of B12.

Since this was 2001, the internet was new and not readily available. I had friends do research on MMA and everything in print was extremely negative. Whatever we read said that children either died in early infancy or had significant developmental delays and constant hospitalizations. We were distraught and stressed about what the future would bring. However, we knew we were lucky that he went into crisis while still in the hospital. All the doctors have told us that if Connor was home we would not have noticed the signs of his crisis until it was too late and would have probably thought it was SIDS.

Connor had every specialist under the sun come and see him – cardiology, orthopedics, neurology. His EEG was abnormal. He had bruises and collapsing veins from his IV's which were in every extremity. I was unable to feed him but told to pump every 3 hours and freeze the breast milk. Eventually, Connor's lab values began to stabilize and improve. Things were looking up. After day 10 we were finally able to hold him and start feeding him a bottle with Propimex-1 and ProPhree mixed with breast milk.

We brought him home after 2 weeks and a nurse came to the house and taught me how to do the shots. Connor was fed every 3 hours round the clock for the first year of his life. We had to set our alarm in the middle of the night and force feed him the bottle, as he just wanted to sleep all night and had little interest in eating. After feeding him, I would pump and save the breast milk, then go to sleep before repeating the cycle again an hour and a half later. Needless to say, I was sleep deprived and delirious, but Connor was growing and doing well.

The first year of his life was definitely the hardest. Constant appointments, round the clock feedings with a baby who didn't want to eat, food diaries, and always holding our breath waiting for the weekly lab results. We also had the worry of negative consequences from the high ammonia levels during his crisis. Luckily we had a great metabolic doctor and nutritionist who would return our phone calls immediately and answer our questions no matter what time of the day or night. When Connor got a fever or a stomach bug we would take him to the ER for IV hydration to prevent a crisis. At first we were given a hard time by the local hospital since they did not know anything about MMA and didn't understand why we were requesting IV fluids for a baby who was not dehydrated or lethargic. But Dr. Slonim told us to go no matter what, and looking back I am so glad that we did because Connor has never had another metabolic crisis since his initial one right after birth. We made it through the first year and Connor met all his milestones on time. His appetite also improved slightly which made the feedings easier.

When Connor was a year old I found out I was pregnant again. I was a nervous wreck throughout my pregnancy. Our second son Aidan was born July 21, 2003 and when he was born we paid to have his blood sample taken and tested for MMA since NY State did not have newborn screening at that time. He is unaffected. My daughter Julia was born June 10, 2005. By then NY had newborn screening and she is also unaffected.

Connor has had his share of childhood illnesses and injuries. When he was 5 he had appendicitis and had his appendix removed but he remained metabolically stable. He has had stitches twice from hockey injuries and surgery to repair a broken finger and nail bed. He has also had many ear infections, strep throat, and a few years ago he had the flu. All of his illnesses were managed from home.

Connor is now almost 11 years old and entering 5th grade in September. He is an extremely active boy. He plays ice hockey (and is a fanatical NY Ranger fan). He also plays lacrosse, soccer, and will be testing for his black belt in karate in October. Connor does well in school and is very studious, but also loves to socialize with his friends.

He gets the B12 (hydroxycobalamin) shot one time per week and takes Carnitor 4cc, 3 times per day. He also gets 75 grams of Propimex-2 per day, but we are in discussions to possibly eliminate the Propimex-2 after going to NIH this past spring. We do not count grams of protein per day but limit his protein intake with a vegetarian diet. He has a great appetite. He loves pasta, rice, veggies, pizza with half the cheese taken off, vegetable sushi rolls, and his new favorite – a sandwich with lettuce, tomato, cucumbers and Russian dressing. Connor would eat junk food all day if we let him and drinks only water unless we let him drink soda. He definitely prefers salty foods over sweet. We see Dr. Slonim twice per year now for a metabolic work up.

I have wanted to share his story so many times, but never got around to it. As everyone knows, life is so busy! I would be happy to talk to anyone in person to share our experiences with MMA. Please feel free to contact me.

by: *Liz , Cold Spring Harbor, NY*
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FOR IMMEDIATE RELEASE

A New Organic Acidemia called "Methylmalonic Aciduria & Homocystinuria, cblJ Type" (cblJ-Hcy-MMA)

David S. Rosenblatt, MD and Dodd Q. Chu and Family Chair in Medical Genetics
Chairman, Department of Human Genetics Professor, Departments of Human Genetics, Medicine, Pediatrics & Biology at Gill University

Vitamin B12 deficiency: tracking the genetic causes

Vitamin B12 is essential to human health. However, some people have inherited conditions that leave them unable to process vitamin B12. As a result they are prone to serious health problems, including developmental delay, psychosis, stroke and dementia. An international research team recently discovered a new genetic disease related to vitamin B12 deficiency by identifying a gene that is vital to the transport of vitamin into the cells of the body. This discovery will help doctors better diagnose this rare genetic disorder and open the door to new treatments. The findings are published in the journal Nature Genetics.

"We found that a second transport protein was involved in the uptake of the vitamin into the cells, thus providing evidence of another cause of hereditary vitamin B12 deficiency", said Dr. David Rosenblatt, one of the study's co-authors, scientist in medical genetics and genomics at the Research Institute of the McGill University Health Centre (RI MUHC) and Dodd Q. Chu and Family Chair in Medical Genetics and the Chair of the Department of Human Genetics at McGill University. "It is also the first description of a new genetic disease associated with how vitamin B12 is handled by the body".

These results build on previous research by the same team from the RI MUHC and McGill University, with their colleagues in Switzerland, Germany and the United States. In previous work, the researchers discovered that vitamin B12 enters our cells with help from of a specific transport protein. In this study, they were working independently with two patients showing symptoms of the cblF gene defect of vitamin B12 metabolism but without an actual defect in this gene. Their work led to the discovery of a new gene, ABCD4, associated with the transport of B12 and responsible for a new disease called cblJ combined homocystinuria and methylmalonic aciduria (cblJ-Hcy-MMA).

Using next generation sequencing of the patients' genetic information, the scientists identified two mutations in the same ABCD4 gene, in both patients. "We were also able to compensate for the genetic mutation by adding an intact ABCD4 protein to the patients' cells, thus allowing the vitamin to be properly integrated into the cells," explained Dr. Matthias Baumgartner, senior author of the study and a Professor of metabolic diseases at Zurich's University Children's Hospital.

Vitamin B12, or cobalamin, is essential for healthy functioning of the human nervous system and red blood cell synthesis. Unable to produce the vitamin itself, the human body has to obtain it from animal-based foods such as milk products, eggs, red meat, chicken, fish, and shellfish – or vitamin supplements. Vitamin B12 is not found in vegetables.

"This discovery will lead to the early diagnosis of this serious genetic disorder and has given us new paths to explore treatment options. It also helps explain how vitamin B12 functions in the body, even for those without the disorder," said Dr. Rosenblatt who is the director of one of only two referral laboratories in the world for patients suspected of having this genetic inability to absorb vitamin B12. Dr. Rosenblatt points out that the study of patients with rare diseases is essential to the advancement of our knowledge of human biology

Funding

This work was funded by the Canadian Institutes of Health Research (CIHR) and by the Swiss National Science Foundation and by the Deutsche Forschungsgemeinschaft.

Research partners

The study was co-authored by David Coelho (first co-author), Terttu Suormala and Brian Fowler of the University Children's Hospitals in Basel and Zürich, Switzerland; David Rosenblatt of McGill and the RI MUHC and his graduate student Jaeseung C Kim (first co-author) of McGill, Isabelle R Miousse, Stephen Fung, David Watkins, Eric A Shoubridge of McGill University and Jacek Majewski of the McGill University and Genome Quebec Innovation Centre; Patricie Burda, Michele Frapolli of University Children's Hospital, Zürich, Switzerland; Martin Stucki and Matthias R Baumgartner of University Children's Hospital and University of Zürich; Marcel du Moulin, Insa Buers, Frank Rutsch of University Children's Hospital, Münster, Germany; Peter Nürnberg and Holger Thiele of University of Cologne, Germany; Nicola Longo of University of Utah, Salt Lake City, USA and ARUP Laboratories, Salt Lake; Marzia Pasquali1 of ARUP Laboratories and University of Utah, Salt Lake City, Utah, USA; Horst Robenek of Leibniz Institute for Arteriosclerosis Research, Münster University, Germany; Wolfgang Höhne of Charité University Medicine, Berlin, Germany; Eugen Mengel of University Children's Hospital, Mainz, Germany.

(*http://publications.mcgill.ca/humangenetics/2012/08/23/in-memory-of-bartha-knoppers-boon/*)

Rayan & Yosef : Propionic Acidemia | 2 1/2 years old

It was the summer of 2009, our son was turning 2 and I found out I was expecting again. My husband and I were thrilled and our biggest concern at the time was making room for one more in our 2 bedroom townhome. Our excitement and nervousness doubled as we found out our new baby was two new babies- we were having twins. My doctor's appointments and ultra sounds multiplied. At around 6 months of pregnancy, it seemed like the odds of an early birth was high, and I was placed on bed rest. It was a tough time as my husband had to travel for work and I had my active 2 year old to care for.

My first son had been born after an emergency C-section, so it followed that my twins had to be delivered the same way. At 2am on February 15th, 2010 I started feeling contractions and knew it was time. I was just over 36 weeks along at the time. My mom had arrived already to help and my sister was planning to fly in as soon as the babies arrived- we needed all the help we could get! I lay in bed timing my contractions while searching for the best flight deals on my phone. By 4am we were on our way to the hospital in Boca Raton, and my sister was packing her things in Chicago.

I was much calmer than I thought I would be. My babies were coming and we were as ready as could be. My ob arrived and everything happened so quickly. At 6.20am my identical twins were born, a minute apart. 2 healthy boys weighing 6lbs04 and 6lbs 01. Rayan and Yosef. My husband was with me, and he was having a great time holding the boys and getting his picture taken with them while I was straining to get a good look. I remember thinking this was one of the happiest days of my life.

Things changed. 6 hours in the room and they had yet to bring me my boys. I was worried about feeding them. What was taking so long? I was told that their blood sugar was high so they had been fed formula. They were currently under heat lamps. I was slightly annoyed as I had not wanted my babies to be fed formula. I was more anxious to get a chance to really meet and connect with the newest members of our family. They finally arrived in the room, bundled up and out cold. Not a peep. This was different. My first had been bright eyed and bushy tailed from the instant he was born. "Spirited little guy" the nurses had said. Everyone told me this is how newborns really are- mostly sleeping. My first born was the different one. Twins were not going to be as hard as I had imagined I thought to myself.

The next day, after the twins had been whipped back to the nursery many times for low body temperature, I was starting to feel nervous. They were twins after all, and born a little earlier than normal. One nurse told me to unwrap them and hold close to my body for warmth. Another yelled at me for doing just that. I was hardly able to feed them. Then to add to it, an ob arrived to circumcise them. We had requested this before their birth, but I felt they were not strong enough. She called me in my room and asserted that they will be fine; it wasn't anything to worry about. I hadn't felt this way for my first but with these two, I was uncomfortable. Still, I thought, the doctor knows what they are going through so if she thinks it's ok, it should be fine. After I gave my ok, I cried. It's my hormones, I told myself.

Later that day, things started going wrong. Rayan was being checked into NICU. How can it be? I cried a lot. I had hardly spent any time with them. They were so healthy when they were born, how could this be possible? Shortly after I was told Yosef was following. It will only be for a couple of days the doc told us. They were just slightly premature. I was going to be leaving the next day- it meant I wouldn't be able to take them home with me. I was a mess.

We came back to visit the next day after discharge. The nicu had strict visiting hours. The twins had feeding tubes in and were hooked up to an IV. It was hard to see them lying so lifeless. The nurses kept asking me if I was sure I was 36 weeks when they were born. They seemed more premature- maybe I had the dates wrong, she suggested. It didn't make sense. But it was ok; these were all typical signs premature babies had.

On day 5, we were visiting our babies again. My phone was on silent while I was inside. We stepped outside to go home and my husband went to bring the car as I was still in pain from my surgery. I saw several missed calls from the same Miami number on my phone and many voicemail messages also. I started listening to the messages in order and suddenly the world stopped making sense. This woman was saying something about my twins having a medical problem. Who was she and why did someone in Miami know anything about my boys. What in the world was newborn screening? And then a message that she couldn't find us so she was having police sent to our house. What? My husband pulled up and saw the look on my face. I was frantically dialing the Miami number. I spoke to the doctor who had been trying to reach me, and she sounded relieved to hear that the twins were in nicu. She hung up to stop the police from going to my house and called me back.

[next page...]

[Rayan & Yosef...]

Those minutes were gut wrenching. She called back and explained to me that my boys had tested positive for a metabolic disorder.

What does that mean? And what could happen to them? She told me on the phone that my twins can go into a coma and die. She hung up to speak to the neonatologist and in a daze we re-parked the car and stumbled into the nicu. We were asked to sit in the conference room as we waited for the doctor to talk to us. Both my husband and I were feverously looking up 'metabolic disorders' on our phones, tears streaming. Just 5 days after the best day of my life, came The Worst Day. I kept hearing the words in my head- 'they can go into a coma and die'. I was picturing the crib in our room that the twins were to share, and all I could think was that it would stay empty.

The doctor came in. He looked upset and he tried to explain to us what it all meant. They still didn't know what kind of metabolic disorder. They wanted to move our babies to Miami, a.s.a.p. He said there was a chance that the test was a false positive. Their ammonia level had come back in the 100s- which he said wasn't too high for children with metabolic disorders. We wanted to believe him….but how could they mess up 2 results?

The next day, early morning on a Sunday, we received a call from the hospital that the helicopter was on its way to transport the boys. We rushed out to the hospital. Only one parent could ride with the babies, and that was to be me. They fit both babies in one incubator and wheeled them out. The nurses hugged me and told me they would pray for them as they handed me a box of tissues for the journey. My first helicopter ride- I had always wanted to see a city from above, but my eyes were on my treasures in the clear box, not the view outside. We arrived at Miami children's hospital and a wheelchair was waiting for me as I could not have kept up in my post-surgical condition. We were wheeled to the nicu, where Dr. Parul Jayakar was waiting for me on a Sunday morning. Little did I know that was the least of her dedication- a specialist working on a Sunday morning. As the twins were taken inside, she started asking me a lot of questions about my pregnancy and also explaining to me what this was all about. And that began our journey as the parents of babies with a metabolic disorder. Our twins stayed in the NICU for 1 month and we would visit every day. During this time we found out that they had a type of organic acid disorder called propionic acidemia (PA). Summarizing some excerpts to try to explain briefly what this means: Organic acid disorders (OAs) are a group of rare inherited conditions caused by enzymes that do not work properly. A number of enzymes are needed to process protein from the food we eat for use by the body and problems with one or more of these enzymes can cause an organic acid disorder. People with organic acid disorders cannot break down protein properly which causes harmful substances to build up in their blood and urine. These substances can affect health, growth and learning. PA causes episodes of illness called metabolic crises, signs of which are poor appetite, vomiting, extreme sleepiness or lack of energy and low muscle tone (floppy muscles and joints.)

Feb: *Both sick again. Yosef developed pneumonia and was admitted to ICU. Rayan was kept overnight with an IV for observation. They were in the hospital for their first birthday.*
March: *Yosef had his first crisis and was in ICU for several days, following a second helicopter ride. He had a cat scan done to check for brain swelling.*
April: *Stomach virus got the best of our whole family. Trip to ER.*
May: *Yosef was back in the ER after falling off the couch and fracturing his wrist.*
June: *All kids severely sick with congestion and ear infections. Many trips to ER. Yosef had high ammonia but continued eating fine.*
July: *Rayan admitted in hospital for constipation.*
August: *Yosef hospitalized for not eating and going into crisis.*
September: *Yosef in hospital for G-tube surgery.*
October: *Finally a month where everyone was fine.*
November: *Rayan hospitalized for constipation.*
December: *Rayan in hospital for constipation again- this time he ended up with g-tube surgery and being in hospital for 10 days.*

They had a genetic test done to see the exact mutation, which determined that theirs was not a mild form. They also had several blood tests done to determine which formula recipe was working best for them. They were still having trouble eating by themselves so they were on an NG tube for a lot of the time. Even after the NG tube was removed, they would take a very long time drinking just 1 ounce of formula. We were taught how to prepare their special formula by the dietician and also by the technician who made it in the formula room. The boys would need this formula for life, adjusted in quantity and composition based on their weight and also by the results of regular blood tests in future.

Finally we were able to take our babies home. They had heart monitors that came with them, which were to be connected at night. We stuck to the hospital routine of feeding them every 3 hours on the dot. They were never hungry and would never cry to be fed so during the night we had to set alarms for every three hours.

It would take an hour to feed each baby, so those were very tough days. My older son was 2.5 and we didn't want to send him to school in case he brought home a virus, so I took care of him also. My mother stayed for 3 months to help out, which was a blessing. We didn't go out anywhere, nor did we let anyone come and visit for several months to keep the twins from getting sick. We also pushed back their vaccines as we weren't sure how their fragile bodies would react, especially if they were to have a fever. Through early steps, we were able to have therapy for our children starting at a very young age. A physical therapist came to our house once a week from the time they were 5 months old. She taught them to roll, and helped to straighten their necks. They had torticollis, a condition twins can have where their neck is not straight due to cramped conditions in utero. At 10 months old, my previously limp and tiny babies started crawling. They were doing great in terms of drinking all their formula by mouth, eating baby food and weighed a healthy 23lbs. They had also never been sick at that point. We had started their vaccines by then and everything was going smoothly. It seemed like they had no health problems and our lives would be 'normal.'

My older son had turned 3 years old and we started sending him to school part-time. It was nice for him to get out of the house and play with friends his age. However, he did start getting sick a lot. And every time he got a cold, the twins would catch it also- only theirs lasted longer and they got much sicker. We made it through their first fevers and breathed a sigh of relief. However, little did we know what 2011 would bring.

January 2011- Yosef and Rayan were extremely sick with a cold and it progressed to bronchiolitis. We were able to avoid hospitalization but they were given a nebulizer and required regular breathing treatments to get better. Every trip to the hospital has a devastating effect on our family. There is the stress, the reality of the disorder- we may lose one or both of our kids, as well as the upheaval of daily life. Through every hospital stay, and even the ER trips, our metabolic specialist Dr. Jayakar was our pillar of strength. She came to see us at the hospital every day, called several times and constantly emailed, not to mention reassure us. During one hospitalization she was out of the country but I still received emails from her around the clock and she was in constant touch with the medical team.

Finally, after the g-tubes were put in, we breathed a sigh of relief. This really cut down on our hospital trips as we could much more easily control their calorie intake, their hydration, as well as the constipation problems. There was more freedom of going out as a family without worrying that they may fall asleep without drinking their formula. When they had colds and couldn't drink due to stuffy noses, it was ok.

We started 2012 with a positive outlook. Our twins had just started to walk, at 22 months of age. Despite going through everything they went through, they were so strong and such happy babies. Then in late January, we discovered something else. They had an abnormal EEG and could be having silent seizures. In February, they were hospitalized for 2 days to do a video EEG and try to determine if they were indeed having seizures. In a period of 24 hours, both had 2 seizures each. They were placed on seizure medication and thankfully, their seizures have stopped, confirmed by normal EEGs in April.

Now Yosef and Rayan are 2.5 years old. They regularly go to a public playground and their immune system is holding up (knock on wood!). Of course, we use a lot of hand sanitizer every time we go anywhere. When we arrive home they also run to the bathroom door (safety locked from outside) and say "open! Hands!" and step up onto their step stools by themselves to wash their hands. They are getting better at climbing up stairs with assistance- and pros at going down a slide! We had been more worried about Yosef as he is a little more behind cognitively than his brother, and not very social. However, since starting the seizure medication he has come a long way. He is making good eye contact and his vocabulary is increasing. He loves numbers and rocket ships. One day at a grocery store aisle he pointed to the number above and correctly said 'eight'. The lady behind us then pointed to the aisle number next to us and asked him what that said- and he told her: "seben". How proud was I that day!

They are extremely opposite in personality and Rayan will pull our neighbors into the house from the door saying 'come on!' while Yosef will push them back towards the door saying 'bye!'. I think having speech and physical therapy from such an early age has helped tremendously. We still have many eating issues with solids but they love certain fruit, can eat baby food without trouble, and drink most of their formula by mouth. We feel very blessed to have come so far. I often think about their lifeless bodies at birth, hooked up to tubes. Then I see my 2 year old Rayan, as he runs up to me, hug me and says "mama! I yuv you!" and every moment becomes worth it. This is a difficult journey, and the future is scary, but if we continue to have more good days than bad, life is great.

by: *Sana and Bilal Tamarac, Florida*
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Propionic Acidemia Moms: July 2012 Portland, OR

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nbs CONNECT

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Mission – NBS Connect will serve as an internet-based support network for parents, guardians and individuals with inborn errors of metabolism. NBS Connect will capture and analyze information related to inborn errors of metabolism in an effort to assess gaps in service, access to care and to develop best standards of practice for clinical management and connect families to research opportunities. NBS Connect strives to improve the quality of care of individuals with inborn errors of metabolism (IEMs).

What is NBS Connect? NBS Connect is a multi-use patient (parent/guardian)-entered data registry. It was designed with the idea of connecting patients and providing information and tools for living with and managing a NBS diagnosed inborn errors of metabolism disorders. In addition, layers of capabilities have also been incorporated for research use. By collecting data on diagnosis, treatment, symptoms, outcomes, barriers to care, and quality of life, we hope to eliminate gaps in service and improve access to care while establishing best standards of practice for clinical management of disease and connecting families to research opportunities.

Who We Are – We are parents, patients, doctors, counselors, administrators, nurses, nutritionists, entrepreneurs, university faculty and patient advocates. Our novel approach to improving the lives of individuals is based on the idea that the most effective healthcare strategy requires a varied group of experts and open communication between patients, academia and industry.

NBS Connect Operational Team:
Kyle Brown, CEO, Patient Crossroads, CA
Rani Singh PhD, RD, LD, Emory University, GA
Yetsa Adadevoh, MPH, Emory University, GA
Sarah Travis, RD, LD, Emory Healthcare, GA

Getting Involved – Registering with NBS Connect and completing a Profile connects you to the latest news and information about inborn errors of metabolism and current clinical trials/research studies and allows you to learn more about the community. Registering also gives you access to information about care and treatment; genetic testing and nutritional services and enables us to tell you about upcoming research studies for which you may qualify.

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Propionic acidemia: To liver transplant or not to liver transplant?

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Propionic acidemia (PA) is an intoxication-type inborn error of metabolism caused by decreased ability of propionyl-CoA carboxylase to convert propionyl-CoA to methylmalonyl-CoA. Affected children typically present with metabolic acidosis, which can evolve to lethargy and eventual coma and death during episodes of catabolism if not adequately treated **(1, 2)**. Recent reports have highlighted the potential for cardiac involvement in PA, especially the development of life-threatening cardiomyopathy, which might be related to abnormalities of mitochondrial function **(3)**. Although propionyl-CoA carboxylase activity is present throughout the body, i.e., not restricted to the liver, success in treating other inborn errors of metabolism such as urea cycle defects, methylmalonic acidemia, and maple syrup urine disease with liver transplantation has made this procedure an attractive therapeutic option **(4, 5)**.

Initial attempts to treat PA by liver transplantation were predominately unsuccessful with high mortality and morbidity **(6, 7)**. With improved surgical techniques and anti-rejection medications and protocols, more recent attempts to treat patients with PA by liver transplantation have been more successful. Many, but not all, patients are able to stop dietary measures, and there is a decrease in number and intensity of acute decompensations, improved cardiac function, and stabilization of neurologic complications. In a multi-site, retrospective study of 12 individuals with PA receiving orthotopic liver transplant (OLT), the one-yr survival rate was 72.2% and there was clear clinical improvement, including the elimination of episodes of hyperammonemia and liberalization of diet (i.e., avoidance of meats but no specific dietary restriction of protein), slowed neurological decline, and the prevention of cardiomyopathy **(8)**.

A recent report described five individuals with median follow-up of 7.3 yr; all were surviving and had good graft function; though, one had experienced a basal ganglia metabolic stroke approximately one yr after transplantation. Three received left lateral segments (one from a living carrier relative), one received a left lobe, and the other had auxiliary partial orthotopic liver transplantation. Post-transplantation development was normal in one patient, moderately impaired in another, and mildly impaired for the other three **(9)**. Another report of three children with PA who received living-related donor grafts (from heterozygous carrier parents) documented survival for 21–59 months following surgery without further episodes of acute decompensation or metabolic acidosis **(10)**.

Kasahara and colleagues in the May issue of Pediatric Transplantation provide further evidence to support the use of liver transplantation in patients with PA, which is present in approximately one in 465 000 live births in Japan. Three children who received living donor grafts between ages seven and 26 months showed stabilization of their clinical disease in terms of neurologic and cardiac findings, and decreased episodes of metabolic acidosis. One patient did have a metabolic decompensation, which was treated using standard methods. Normal liver graft and cardiac function, as well as normal development and decreased hospitalization were also described in these children. The fact that liver transplantation does not resolve the underlying biochemical defect completely was clearly demonstrated by the persistence of pathognomonic PA metabolites in blood and urine postoperatively.

In January 2011, a group of clinicians, scientists, and patient representative groups provided a number of acute and chronic health management guidelines for patients who have PA. On the basis of most recent reports of positive outcomes in liver transplantation in patients with PA, consideration of liver transplantation in individuals with recurrent episodes of hyperammonemia or acidosis that are not adequately controlled with medical therapies is among the recommendations **(11)**. Moreover, findings seem to support that the recipients of living-related donor livers from carriers (i.e., haploinsufficient livers) seem to have similar results to OLT recipients, and the current work by Kasahara and colleagues supports this finding **(11)**. Liver transplantation may soon be considered the treatment of choice for PA; given the positive clinical outcome data and continued refinements in surgical and post-operative techniques, the risk-benefit ratio continues to improve. Nevertheless, questions related to optimal post-operative management, long-term effects on cardiac function and developmental outcome remain, so further experience and careful longitudinal follow-up of transplanted patients will be required.

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Memorial

Charlene, Glutaric Acidemia, Type 1 – July 11, 1992 - May 16, 2012

My angel was called home on May 16th, 2012. She passed peacefully in her sleep after 19 years of fighting. Charlene was diagnosed with Glutaric Acidemia Type 1 in April of 1993.

Charlene loved family and friends. She was always smiling, even during sick times. Char was an adventurist! She loved going on rides at the fair, roller coasters were her favorite!! She was a teacher. She taught everyone who knew her that life is what we make of it. She didn't let her disability hold her back. She taught us how to smile at everything, how to laugh at the silly things, how to love with all we have, and how to have faith in God. Char stayed active; playing baseball, going to church, shopping, theme parks, boardwalk, school, and she was even on the Today Show once (she did the weather with Al Roker)! Char was the center of our family. We do not say goodbye, we will see you someday my darling. You are and always will be Mommy's angel, my heart and my soul.

Advocate for Your Child

All parents advocate for their children as they speak up for them and help them to be successful. When your child has a serious health condition or a disability, you have more opportunities to advocate for your child because your child faces many challenges. You and your child will need to work with many people and organizations. You will be your child's best advocate.

Raising a child with a serious health problem or a disability can be like training for and running a marathon. It will take a long time to reach your goal. It will require a lot of work. You will be very proud when you reach your goal with your child. Successful parent advocates plan ahead, train, work with coaches, and develop winning strategies. They help their children grow and develop in spite of the challenges.

Here are some things you can do to be a good advocate for your child:

Take care of yourself.

- Rest, eat well, and take breaks so you can stay strong.
- Let others help you. Family and friends want to help. Tell them what you need.
- Pay attention to your feelings. Get support when you feel sad.

Become an expert on your own child.

- Don't be afraid to speak up.
- Look people in the eye when you talk to them.
- Ask them to explain again, when you don't understand.

Learn about your child's condition.

- Ask questions at clinic visits. Write them down ahead of time. Bring someone to take notes for you.
- Save all the information you get. Keep it in a notebook or a file cabinet.
- Be careful of what you read on the Internet. It is not all true. Check it out.

Learn where your child can get help.

- See if your child can get help from your school district.
- Babies who are delayed can get free help from early intervention. Call your school district, HeadStart Program, or county health and human services.
- Early intervention and special education in school are rights for children with special needs. Learn about these rights to help your child.

Help your child be independent.

- Plan for your child to succeed and grow up strong.
- Teach your child about the health problem or disability.
- Show your child how to be a good advocate
- Introduce your child to others with the same condition
- Provide good role models

Keep track of contacts.

- When you talk to people on the phone, write down their names and numbers in a notebook.
- After the call, write notes before you forget. For example, "Mai Vang 651-555-1234 said she would mail a form for TEFRA." If the form doesn't come, you know who to call.
- Make important requests in writing and keep a copy.

Let healthcare providers know if you can't pay for something.

- If you have insurance, learn what it covers.
- Ask for help if payment is denied
- Check to see if there are programs to help pay for your child's needs.
- Save all the paperwork and bills you get.

Talk to other parents.

- Get to know parents whose children have the same problem as yours.
- Join a parent support group
- Talk to other parents whose children have different problems than yours. Ask for hints. Learn from them
- Contact a parent-training center. There is one in every state Find yours at *www.parentcenternetwork.org/parentcenterlisting.html* You might not need them now. Their ideas will help you in the future

by: *Beth-Ann Bloom, MS, CGC Genetic Counselor. Beth-Ann is the parent of a young adult with a disability and has enjoyed over 30 years of working with families dealing with metabolic and other genetic disorders*

New insights into cognition and learning in patients with isolated MMA

Jennifer Sloan PhD MS, Irini Manoli MD PhD, Joseph Snow PhD and Charles P. Venditti MD, PhD

Since our last article update, we continue to study hereditary methylmalonic acidemia and cobalamin disorders in both the laboratory and the clinic and are extremely grateful for the families that have taken the time to participate in our study at NIH. This article describes a research study that we published this year, which is a direct result of families coming to the NIH and participating in standardized neurocognitive testing.

When parents are given a diagnosis of MMA, they are often told that their child may have developmental delays, learning problems or intellectual disability. Parents may think: How will my child do in school? Will they be able to work or live independently? Unfortunately these questions are difficult to answer because each patient is unique and also because there is not much written in the medical literature about the cognitive outcomes in children and adults with MMA. In our recent paper published in Pediatrics in May 2012 [reference: O'Shea, C.J. et al. Neurocognitive Phenotype of Isolated Methylmalonic Acidemia. Pediatrics 129(6):e1541-51 (2012)], we studied the cognitive and neurological status of 43 patients, ages 2-32 years, with isolated MMA (mut, cblA, cblB) over a period of 6 years. Most of the patients studied (40/43) were diagnosed with MMA after they developed symptoms.

To understand our study it is helpful to learn about intelligent quotient (IQ) scores. The average IQ in the general population is 100, with 90-109 considered average, 80-89 low average, and 70-79 borderline. People with an IQ under 70 are considered to have an intellectual disability if they also have deficits in adaptive functioning. Our data showed that the IQ scores of people with MMA vary quite a bit, ranging from ~45 to ~120. The average IQ for all MMA patients was around 85, which is in the low average intelligence range. Although the IQ scores tended to be lower in MMA patients than in the general population, IQs were stable in patients who were tested over several years of the study. This suggests that cognitive decline is not necessarily a feature of the disease.

Patients that had mut MMA and were diagnosed in the first month of life had average IQ scores of ~70 compared to patients with cblA and cblB, who had average scores of ~100. At the time of diagnosis, more than half of the patients in the study had an excess of ammonia in the blood that can injure the brain. Our analysis showed that elevated ammonia also correlated with lower overall IQ, as well as verbal comprehension and perceptual reasoning/organization. We also found that on neurocognitive tests, people with MMA do appear to have a particular deficit in processing speed, which involves fluently performing simple tasks. This may reflect changes to the part of the brain known as the basal ganglia, which we know has been damaged in patients with MMA who have had a "metabolic stroke".

Our study establishes a valuable set of data and framework for future investigations such as comparison to a group of younger patients born after the generalized implementation of newborn screening. We are hopeful that earlier diagnosis by newborn screening, especially of the patients with later onset, and new medicines/treatments for MMA will help to prevent complications like recurrent high ammonia levels, metabolic stroke in the basal ganglia and will improve the overall IQ scores in patients with MMA in the future. Understanding the cognitive outcomes and specific challenges for patients with MMA are key to providing optimal care for those with MMA.

We would like to thank all of the patients and families who participated in our study over the years. Our partnership is essential to increase the knowledge about MMA and cobalamin disorders and we are certain it will continue to advance the field. If you are interested in participating in our study you can find out more information here: *http://www.genome.gov/19016897*

You can download the paper here: *http://www.ncbi.nlm.nih.gov/pubmed/22614770*

Colorado Families meet and greet in Denver, CO May 2012

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ANNUAL DONATION CHANGE OF ADDRESS

Please accept $_____ as our annual tax deductible donation to the Organic Acidemia Association.

Suggested membership donation is $25 (US) and $35 (international). Extra funds are welcome and can be designated for research, OAA operating expenses, or to help others attend the Atlanta Conference.

Name: _____

Address: _____

City State Zip: _____

Email: _____

Please make the following changes to my address, phone number, or email address.

Remember the newsletter does not get forwarded when you move!

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.

- The information contained herein does not necessarily represent the opinions of our Board of Medical Advisors or Board of Directors
- Letters and photographs sent to OAA become the property of OAA and may be used or edited at the discretion of the OAA staff.
- Names or information will be kept confidential only if specifically requested in writing.
- This newsletter does not provide medical advice. You should notify your health care provider before making treatment changes.

Find us on Facebook

OAA is on Facebook - donations can be sent through our "Cause" Page, connection with other parents can be found through our private "OAA Group" and private "Fan" Page.

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.