

# Propionic Acidemia Foundation

VOLUME 1, ISSUE 24

SPRING 2018

## PAF Exhibits at SIMD

Jill Chertow represented PAF and the PA Community at the 40th Annual Society of Inherited Metabolic Disorders (SIMD) on March 11-14th in San Diego, California. PAF partnered with the National Urea Cycle Disorders Foundation (NUCDF) on our exhibit booth to educate providers about hyperammonemia.

PAF's goal was also to network with PAF clinicians and attract researchers to collaborate with PAF on studies for PA. The meeting was a great opportunity to hear about new research in metabolic disorders, newborn screening updates and issues about access and cost of drug therapies. Elaina Jurecki, M.S., R.D. presented on "Protein Intake Recommendations for Propionic Acidemia in the Evidence-based SERN-GMDI Management Guidelines." Nutrition guidelines continue to be a hot topic. Interesting poster topics included the use of medical food, liver transplant, anaplerotic therapy and an update on the PA study at NIH.



During the breaks and meals there was time to network with physicians, dietitians, researchers, medical food providers, advocacy groups, and industry. There is hope for a number of potential new therapies in mRNA, gene therapy, enzyme replacement therapy and/or new medications.

### SAVE THE WEEKEND

**October 20, 2018– PA Family Day, Columbus, Ohio**

**October 21, 2018– Nationwide Columbus Marathon & 1/2 Marathon**

**JOIN TEAM PAF/PAR!** Run or cheer to help find a cure for PA, and raise awareness. All running (and fast walking) abilities welcome! You can also participate in a town and distance you enjoy the most. For more information, contact Marisa Cotrina, TeamPAR4@gmail.com.

### PA Registry

Help move research forward for propionic acidemia. Participate in the Propionic Acidemia International Registry. As of March 1st, there are 60 participants. For more information on joining the registry, or to update your information, go to [www.paregistry.org](http://www.paregistry.org).

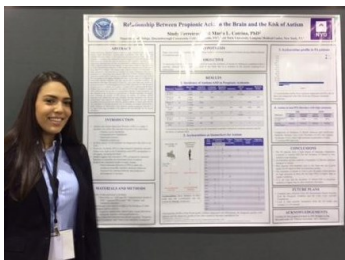
### INSIDE

PAF RESEARCH	2
ANNABELLE'S STORY	3
NILA'S STORY	4
LIVER TRANSPLANT FAQ	5-7
PAF MRI STUDY	8
MINDFULNESS MEDITATION	9
PAF EVENT & FUND- RAISING SPOTLIGHT	10
BIRTHDAY WARRIORS PARTNERSHIP	10

**MISSION:** The Propionic Acidemia Foundation is dedicated to finding improved treatments and a cure for Propionic Acidemia by funding research and providing information and support to families and medical professionals.

**VISION:** To create a future where Propionic Acidemia can be prevented and any affected individual can be cured and live a productive life.

## PAF SPONSORS RESEARCH ON PA BY MINORITY STUDENTS



Last summer, Propionic Acidemia Foundation (PAF) established a collaboration with Dr. Patricia Schneider from the department of Biology at Queensborough Community College

(QCC, Queens, New York) to sponsor a project on the impact of propionic acid in the incidence of autism in Propionic Acidemia (PA) affected individuals. The project was part of the research initiative “Bridges to the Baccalaureate”, a National Institute of Health (NIH) funded project that provides resources for a summer research

project for minority students. Designed and mentored by Dr. Marisa Cotrina, herself the mother of a PA child, this work investigated the incidence of autism in the propionic acidemia population and the validity of mouse models of autism to study the impact of propionic acid in brain. A unique asset of the project was the utilization of the data collected by the PAF PA International Patient Registry. The authors of the study are currently preparing a manuscript for publication of the results found.

At the end of the project, our student, Sindy Ferreiras, had the opportunity to present her research in the area of Neuroscience at the Annual Biomedical Research Conference for minority students (ABRCAMS) that took place in Phoenix, Arizona last November. Well done, Sindy!

## MANUSCRIPT SUMMARY OF PAF FUNDED PROJECT

by Marisa Cotrina, PhD

*Title: Import of TAT-conjugated propionyl-CoA carboxylase using models of propionic acidemia.*

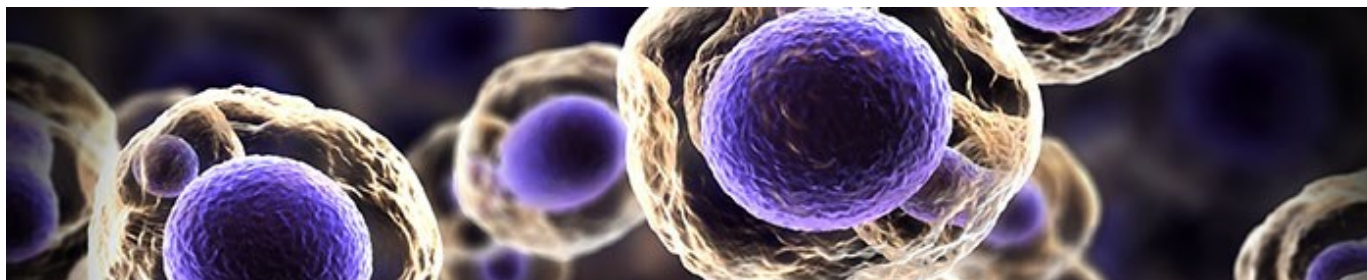
*Authors: Renata Collard, Tomas Majtan, Insun Park and Jan P. Kraus, Department of Pediatrics, University of Colorado School of Medicine, Aurora, CO 80045, USA*

*Journal: Molecular and Cellular Biology – January 2018*

Summary of the work: A new publication came out last month about a possible strategy to introduce “healthy”, non-mutated propionyl-CoA carboxylase (PCC) into cells of patients with propionic acidemia, where PCC is mutated, non-functional. The challenge that Dr. Kraus and colleagues engaged in was to test whether a technique using a “TAT peptide” transporter would work as “enzyme replacement therapy” with a protein of the size and complexity of PCC, formed by twelve different smaller subunits that need to remain together until they get to their final destination, the mitochondria, and then form the final, fully functional, active enzyme. The key experiments in this work were to prepare in the laboratory a synthetic protein of PCC, attach it to the TAT transporter system and then test if this construction would make it to the mitochondria intact and active. Through a series of beautiful biochemical and cellular

experiments, Dr. Kraus team proved that their technique worked not only to introduce the active protein in isolated liver mitochondria but also in PA-patient derived fibroblasts cells (isolated from the skin). The research team then moved one step further by injecting their TAT-PCC conjugate into a mouse also lacking the PCC enzyme (like PA patients). What they found was that the mice not only showed mitochondria with a functioning PCC protein but that this PCC enzyme was also able to temporarily reduce the levels of abnormal propionyl-carnitine in plasma.

What is the potential of using the TAT technique in PA patients? The success of the experiments described in this report consists in the ability to preserve the shape and activity of a very large protein like PCC, never tested before, during the process of delivering it to its final destination. However, for the technique to be implemented in humans, the PCC protein needs to get into the mitochondria in larger amounts and remain active for longer periods of time than what is achieved here. Although still a premature work to move into clinic, a very exciting approach that we hope will continue to progress into the next phase.



## ANNABELLE'S STORY

Annabelle Grace was born September 20, 2016 and like many children with Propionic Acidemia (PA) she went into metabolic crisis a couple of days after birth. After talking to our genetics team in December 2016 we started pursuing the option of a liver transplant for Annabelle. Even though we managed Annabelle's care so meticulously she would still end up in the hospital every couple of months for high ammonia levels above 100 (often for no reason and with no detected illness). Annabelle had a g-tube placed when she was two weeks old, but even with pushing fluids and using sick day formulas we found it difficult to keep her metabolically stable (we checked her ketones EVERYDAY). We tried Carbaglu (which is supposed to help lower ammonia) along with high doses of Carnitine and Bicitra (Sodium Citrate), and those medications didn't even seem to help control her metabolic instability. Her ammonia on a "good day" seemed to hover in the 60's or 70's, and even the night before her transplant her ammonia was 71. We realized early on that Annabelle had a very severe mutation, and we were told by many medical professionals that she was one bad illness away from another metabolic crisis that could cause serious brain damage.

After talking with other families we were told that the Children's Hospital of Pittsburgh was the place to go for transplant. The only liver transplant option in our state (North Carolina) was Duke University, but they had never performed a liver transplant on a child with PA. In April 2017 we ventured up to Pittsburgh, PA for Annabelle's liver transplant evaluation and immediately fell in love with everything this hospital had to offer. Annabelle was officially listed for transplant on May 2, 2017. We immediately started fundraising and used the assistance of COTA (Children's Organ Transplant Association) which we cannot recommend enough. Within three months our team of volunteers raised over \$50,000 for COTA in Honor of Annabelle to help with any transplant-related expenses.

On August 9, 2017 we got "the call" that would forever change our lives, and we quickly rushed to Pittsburgh. We were very fortunate that our first call was "the call" that gave Annabelle her new liver. She went back for surgery around 10:30pm that night and they finished her surgery around 9am that next morning. After surgery Annabelle spent about one week in the PICU.

After that week the transplant team moved her to the transplant recovery unit where she stayed until she was discharged. Around two weeks post-transplant Annabelle did encounter a small episode of rejection. Even though "rejection" sounds scary it is very common early on in transplant, and mild cases like Annabelle's are generally treated with some high-powered IV steroids for a few days. Annabelle was discharged on August 30th and only spent a

total of 21 days in the hospital. The transplant/genetics teams in Pittsburgh told us to prepare for complications (as is common with Organic Acidemia patients), but overall Annabelle had very few complications from her transplant surgery for which we are thankful.

After getting discharged we were required to stay in the Pittsburgh area until the transplant team decided she was stable enough to return home to North Carolina. Luckily, the Ronald McDonald House there is amazing, and instead of hotel rooms they have small one-bedroom apartments making it possible to live there for an extended period of time. Plus, it is one of the few Ronald McDonald Houses where it is connected to the hospital, so even when Annabelle was inpatient we were able to easily access their services (homemade meals, laundry, therapy pets, etc.). We stayed

in Pittsburgh until late November mainly going to the hospital for weekly labs, therapies, and clinic visits. The team had to keep changing her medications weekly so that her liver numbers and her EBV levels (Epstein-Barr Virus that she acquired from her donor) maintained a healthy balance. Our total stay in Pittsburgh was a little over three months, which we were prepared for since the transplant team told us prior to surgery to expect to stay there anywhere from three to six months depending on the amount of complications.

Since we've been home it has been a bumpy road. The transplant team told us that the first winter post-transplant is always very difficult and they were right! Annabelle has been living in a bubble all winter, but she has still been in and out of the hospital the past few months due to illnesses from her immunosuppression. When we do have to go to the hospital its more for treating the illness caused from her immunosuppression rather than treating her underlying metabolic disorder. When you get a liver transplant it really is just trading out one disease (PA) for the other (transplant) in hopes that treating the transplant gets easier in time.

Even though the liver transplant brings a whole new set of issues (more frequent bloodwork, more meds, life-long immunosuppression), we know that Annabelle is more metabolically stable on a day-to-day basis. Even when she does get sick we don't worry as much about the significant possibility of brain damage because her ammonia levels stay within the normal range or are only slightly elevated. (continued on pg. 11)



## NILA'S STORY

Nila Rechelle was born 1/6/12 at 4:02am via emergency cesarean. She was three weeks early and weighed in at 6lbs 4oz and 19 inches long. She passed all initial screenings and appeared to be a healthy baby girl. She did struggle to latch, so I pumped and bottle fed her. She did not make any attempt to suck whether it be breast or bottle. The first day she was able to keep the breastmilk down, but then I was unable to produce anything, so we introduced formula to keep her fed. Shortly after the first formula, Nila began to projectile vomit anything that went in. Joe and I were concerned for her not being able to keep anything down. The nurses over and over tried reassuring us this was completely normal for a new baby. The next day Nila began to turn jaundice on us. She had to stay under bilirubin lights 24 hours a day and only out of the incubator to feed and diaper changes. Her color was improving minimally and her feeding seemed to improve slightly. She had to stay in the hospital an extra day. The doctor released us and we went home. All seemed well for the first few days home with the exception of a few vomiting episodes. Then, we received a call from our pediatrician letting us know we needed to take her to the hospital the next morning for additional blood spots and urine analysis. He let us know her newborn screening came back with signs of a metabolism disorder similar to PKU. He scheduled her first appointment with Riley Children's Hospital in Indianapolis the following Monday with the Genetics team. The blood and urine were rushed off to Duke and we worried and waited impatiently for Monday. We got to Riley and met the team, they informed us of her diagnosis of Propionic Acidemia. This was a huge blow for us. How could our perfect baby girl have to deal with such a terrible disease? How were we going to manage this? So many questions and so many unknowns. Lucky for us, we came in contact with several families through Facebook forums and the dieticians and genetics counselor became family and we were in constant contact. What seemed so scary at first, motivated us and we learned so much and found out it wasn't as terrible as we thought.

Nila struggled with her initial milestones such as holding her head up, rolling and crawling. She had Physical Therapy, Occupational Therapy, and Developmental Therapy several times each week. Once we strengthened her tiny muscles she took off. She zoomed through her milestones! She walked right before 10 months and then she was released from therapy. She has had 17 hospital stays and many more lengthy ER visits for D10 fluid boosts. Ear infections, gastrointestinal bugs and the periodic cold were the main reasons for her hospital stays and visits. The only metabolic stay was a 1 week stay after she had gotten RSV. We were close to getting a tube in but Nila got

stronger each day with her sick formula and D10 and started eating again. To date, she presents as a mild case of PA, but we do not take that lightly as she may begin to show late onset symptoms as she gets older.

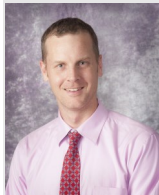
She has always eaten by mouth. She was recently taken off of her metabolic formula and she loves it! She has an extra 8 grams to eat in regular foods in addition to the 28 grams she already gets through food. She enjoys mac-n-cheese, chips, mushrooms, fries, fruits of all kinds, pickles and popsicles galore. Our biggest struggle now is to make sure she gets her daily protein intake goal met and to make sure she is not sneaking food.

Nila is thriving and doing everything a 6 year old girl is able to do! Nila started Kindergarten this past fall and she is one of the top students in her class. She was selected to participate in the High Ability class at one of our local schools. She is smart as a whip and is learning so much in her outdoor kindergarten class. She loves gymnastics, playing softball, riding her bike, fishing, gardening, painting nails and doing her make-up, digging for worms, exploring outdoors and making mud slime!



## LIVER TRANSPLANTATION FOR PROPIONIC ACIDEMIA: FAQ

### PART 1: Answers to Questions that Families May Have



James Squires, MD, MS  
*Dr. Squires is a liver disease specialist at Children's Hospital of Pittsburgh of UPMC and an assistant professor of pediatrics at the University of Pittsburgh School of Medicine.*

Jodie M. Vento, MGC, LCGC

*Jodie Vento is a genetic counselor and manager of the Center for Rare Disease Therapy at the Children's Hospital of Pittsburgh of UPMC.*

#### ***What can we expect that a liver transplant could do for our child?***

Based on experience to date with liver transplants in children with Propionic Acidemia (PA), we can say that after a liver transplant, children are likely to have a substantially better quality of life and a dramatic reduction in metabolic crises. It's important for families to understand, however, that liver transplantation is **not** a cure for PA. *This is because the enzyme deficiency that causes PA exists throughout the body, not just in the liver.*

The liver transplant serves as what we liver specialists call a *bulk enzyme replacement, providing enough functional enzyme to minimize – if not eliminate – metabolic crises, which are the most severe complications of PA for affected children as well as one of the most frightening features of the disease for families.*

Because complications related to PA may still occur following a transplant, there will be a continued need for your child to get follow-up care with one or more medical specialists.

#### ***Is there a minimum or "best" age for a child with PA to have a liver transplant?***

There is no minimum or "best" age. At our center, the average age of a liver transplant for a child with PA is about seven years old, but we have performed transplants in children as young as one year old.

The best time to consider a liver transplant is while the symptoms of PA are still reasonably well controlled. There is also no minimum age for undergoing a pre-transplant evaluation or being placed on the transplant waiting list.

#### ***What should we consider when deciding where to take our child for a liver transplant evaluation?***

The most important factor to consider is the experience of the surgical team performing liver transplants in patients with PA and other metabolic diseases. These patients have complex needs that are different from those of patients receiving liver transplants for other conditions.

The pediatric liver transplantation program at Children's Hospital of Pittsburgh of UPMC was established in 1981 by world-renowned transplant surgeon Thomas E. Starzl, MD, PhD. Our Director of Pediatric Transplantation, George Mazariegos, MD, FACS, pioneered liver transplantation for children with metabolic diseases in 2004. Since that time, Children's Hospital has performed more than 330 liver transplants for children with metabolic disease, more than any other transplant center.

We've also performed more liver transplants in children than any other center in the United States and more living-donor transplants than any other pediatric center in the country. Our one-year survival rate for pediatric liver transplant patients is 98%, exceeding the national average of 95%, according to the Scientific Registry of Transplant Recipients, Jan. 2018 release.

In addition to our world-renowned and experienced liver transplant surgeons, our Center for Rare Disease Therapy includes international experts in the diagnosis and treatment of PA and other metabolic diseases.

#### ***How would we start the process of having our child evaluated for a liver transplant?***

I can tell you how the process works here at Children's Hospital of Pittsburgh of UPMC. It starts with a referral from your doctor or hospital requesting that we evaluate your child. We also receive self-referrals directly from interested families. We will ask the doctor or hospital, or both, to send us all of your child's medical records.

We will look at the records carefully to help us understand your child's medical history and current situation. This information helps our multidisciplinary team develop an individualized plan for your child's evaluation visit. For example, if your child has recently had certain laboratory or imaging tests done, we won't repeat those tests unless there's a valid medical reason for doing so. Understanding how the disease is affecting your child helps us identify which specialists your child should see during the evaluation. (continued next page)

## LIVER TRANSPLANT FAQ (CONTINUED)

It's important for families to know that undergoing a pre-transplant evaluation involves no commitment on either side. It carries no guarantee that your child will be listed for a transplant or, conversely, any requirement that you must agree to have your child placed on the transplant waiting list. We can answer questions, provide information, and make recommendations. Ultimately, however, the decision to proceed with a transplant, or not, is a personal one for each family to make.

The evaluation is an opportunity for the family and the health care team to meet and get to know each other, as well as for the family to gather information and get answers to any and all questions you may have. We hope you'll feel comfortable raising any concerns. Please don't hesitate to ask us about any issue that's on your mind. There are no dumb or silly questions. And, of course, if after you've gone home you think of something that you wish you had asked, please give us a call.

You can expect that the evaluation will be a two- or three-day event. The staff of our Center for Rare Disease Therapy will work with you to arrange for you, your child, and other family members to stay near the hospital, either at our Ronald McDonald house or at a nearby hotel, while you're here for the evaluation.

We'll send you a schedule in advance of your visit. This will tell you which medical and surgical specialists you'll be seeing at what times and what laboratory or imaging tests we would like your child to have during the evaluation. To the extent possible, we try to anticipate all the testing we'll need so that it's a relatively smooth process while you're here.

***Please tell us more about what we can expect during our child's evaluation.***

Because PA is a genetic disease, the specialists you'll see will likely include a medical geneticist and a metabolic dietician. Also, because PA often causes heart problems, your child's evaluation is likely to include basic heart function tests and an assessment by a cardiologist. Depending on how the disease is affecting your child, the evaluation may also include visits with specialists such as the following:

- A neurologist, to assess brain function
- A gastroenterologist, to assess pancreas function
- A hematologist, to assess bone marrow function

Although we try to anticipate all the testing we'll need and schedule it in advance, sometimes we may decide that it would be helpful to do an additional test that wasn't originally on the schedule. For example, depending on the results of the basic heart function tests, the cardiologist might want to do a "stress test" that will provide more detailed information and measurements relating to how well your child's heart is functioning.

***If we decide to go ahead with listing our child for a transplant, what are our options for obtaining a donor liver? How long can we expect it to take to find a compatible donor?***

PA is considered a high-priority condition for liver transplantation, so your child's name will be near the top of the waiting list. However, because demand for donor livers is high and supply is limited, I tell families to be prepared to be on the waiting list for several months.

With any liver transplant, careful testing needs to be done to ensure compatibility of the donor liver and the transplant recipient. Many factors can influence the waiting time for a compatible organ. For example, a child with an uncommon blood type may face a longer wait.

In general, child-size donor livers are scarce. A unique feature of the liver, however, is that it is the only organ in the human body that can regrow. This means that in some cases it's possible to transplant a section of a healthy liver rather than the whole organ. For example, a child who needs a liver transplant may receive a section of a liver from an adult donor. You may hear this type of transplant referred to as a "reduced-size" or "split" liver transplant.

Another type of liver transplant involves a living person – such as a relative, friend, or even a stranger – donating a section of their liver to someone who needs a transplant. Living-donor transplants may be an option for some children with PA. However, because PA is a genetic disease, parents and possibly siblings may be carriers of one of the genetic defects that cause the disease. Someone who is a carrier would not be a suitable living donor.

The good news is that children who receive a partial liver seem to do just as well as those who receive a whole liver. All of the options for obtaining a donor liver, including a reduced-size, split, or living-donor transplant, are discussed during the pre-transplant evaluation. (continued next page)

## LIVER TRANSPLANT FAQ (CONTINUED)

*We've decided that a liver transplant is right for our child. What are the next steps?*

When your child's name is placed on the liver transplant waiting list, we will give you a pager that you will need to take with you everywhere you go so that we can reach you right away when we get a call that a matching donor liver is available. We don't know when that call will come, but when it does you'll need to be able to get to Children's Hospital in a safe, but timely fashion. The transplant team will work with you to establish a 'travel plan' for you and your family for when the transplant is likely to occur.

While your child is on the waiting list, our specialists will work with your local doctors to care for your child and optimize their medical condition ahead of the transplant.

We know that waiting can be a difficult time for families. Your transplant coordinator is always available to respond to your questions and concerns and can also help you make travel arrangements.

Once you arrive at the hospital, preparations for the transplant may take from 12 to 24 hours. Your child will undergo another round of tests to confirm that the donor liver is a good match. Your child will also need to fast before surgery. Our metabolic dietitians will help us prepare intravenous fluids to provide your child with an individualized balance of fats, protein, and glucose to maintain stability while they can't take anything by mouth.

The liver transplantation surgery may take up to several hours, although this varies in each case. While your child is in the operating room, a member of the transplant team will keep you informed on the progress of the transplant.

After the surgery, your child will go to the intensive care unit to be monitored closely until their condition is stable. Then your child will be moved to the liver transplant unit. Staff here will help you learn about your child's medications, diet, need for follow-up care, and anything else you'll need to know to care for your child.



*After the transplant, will our child have to take anti-rejection medication?*

After a liver transplant, you should expect that your child will need to take medication for the rest of his or her life to prevent organ rejection. The body's normal reaction to a transplanted organ is to recognize it as a "foreign agent" and mount an immune response against the new liver. Anti-rejection medications suppress the immune system, which is the body's defense system against illness and infection, to prevent it from attacking the new liver.

Because anti-rejection medications weaken the immune system, your child may be more likely to get infections – and those infections will be harder to treat. You will need to notify the transplant team at the first sign of an infection, such as a fever, chills, sweats, coughing, nasal congestion, diarrhea, redness or swelling, pain, or vomiting. A referral to a doctor may be needed as well.

With immune-suppressing medications, the goal is to find a treatment plan that achieves the needed degree of immune suppression while causing the fewest and least harmful side effects. Regular blood tests will help your child's doctors monitor the medications' effectiveness.

The risk of organ rejection declines over time. This means that in time your child should be able to take lower doses of anti-rejection medications. Most likely, however, he or she will need to continue taking at least a low dose of immune-suppressing medication life-long.

Here at Children's Hospital of Pittsburgh of UPMC and elsewhere, research is underway to learn more about whether some liver transplants patients can eventually stop taking immune-suppressing medication without increasing their risk for rejection of the transplanted organ. This research is a long-term effort, however, and it will be years before we can answer this question.

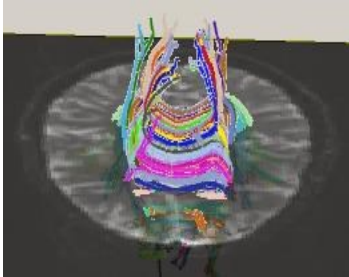
*For more information, please visit: [www.chp.edu/rarecare](http://www.chp.edu/rarecare) or call (412) 692-RARE (7273) In Part 2 of this article, Dr. Squires will summarize the findings of a recent study of outcomes in children with PA and methylmalonic acidemia who received liver transplants at Children's Hospital of Pittsburgh of UPMC.*

The Propionic Acidemia Foundation Newsletter is designed for educational purposes only and is not intended to serve as medical advice. The information provided should not be used for diagnosing or treating a health problem or disease. It is not a substitute for professional care. If you suspect you or your children may have Propionic Acidemia you should consult your health care provider.

The Propionic Acidemia Foundation does not recommend nor endorse any particular products, companies, or manufacturers.

## MRI STUDY AT CNMC AND GEORGETOWN

### Do you have Propionic Acidemia and are between the ages of 6-40 years of age?



**Participate in  
a MRI Study at  
Children's National Medical Center (CNMC)  
and Georgetown**

CNMC and Georgetown are seeking patients with propionic acidemia (PA) between the ages of 6-40 to participate in a study on brain function in PA.

This study will investigate brain function and metabolism in PA relative to healthy control subjects to understand how having PA interferes with cognitive functioning.

This study is being conducted with researchers in Washington, D.C. at CNMC and the Center for Functional and Molecular Imaging (CFMI) at Georgetown University.

#### **What does participation mean?**

- MRI (magnetic resonance imaging) which takes a picture of your brain. It is safe and there are no injections
- You must be able to lie still as we don't use any sedation medication
- You will also be asked to play memory games and complete some cognitive testing
- You will also perform testing while wearing a headband that measures changes in blood flow when you perform a task. This is called functional near infrared spectroscopy (fNIRS) and uses lights to make maps of brain function and is safe and non invasive.

#### **To be eligible to participate, you must:**

- Be between the ages of 6-40 years old
- Have no metal in the body
- Be able to follow written and verbal instructions in English, but English does not have to be your first language
- Be a male or female PA patient

#### **You are not eligible to participate if:**

- You are outside the age ranges listed
- You are pregnant
- You have any metal in your body that is not safe with MRI or have braces or permanent retainers on the teeth which degrades the MRI pictures
- You are claustrophobic and can't have MRI without sedation
- You have a significant history of neuropsychiatric illness or drug use or are metabolically unstable at the time of the study

**Compensation is available: plus parking and local travel**

#### **Contact Information:**

To schedule a phone interview and testing visits, subjects may contact us at either our Washington D.C. Macy Curell 202-687-7823 (study coordinator) [mac518@georgetown.edu](mailto:mac518@georgetown.edu) or Dr. Gropman [agropman@childrensnational.org](mailto:agropman@childrensnational.org)



## MINDFULNESS MEDITATION

*An Ancient Tradition with Practical Application*

Vicki Ancell Sheahan, MBA, CPC, CCC

Pathway Coaching

*"We cannot stop the waves, but we can learn to surf" Jon Kabat-Zin*

Meditation has taken its rightful place in western society. The benefits of meditation, from reducing stress, modulating hormonal functioning, especially of oxytocin and cortisol, and reducing the intensity and frequency of negative and chronic stress reactions are now being documented and used by medical doctors, medical clinics, such as Mayo, and many other professionals today.

Meditation strengthens our ability to cope with difficult emotional experiences and increase emotional wellbeing by mitigating negative thinking, including rumination. Meditation as a way of being teaches us to manage the "narrative" in our head and helps us become emotionally proactive rather than reactive.

Mindfulness Meditation is a method that focuses on our breathing, noting when our mind wanders, and gently returning our attention back to our breath. This focus on our breath, noting our wandering mind and returning to our breath is training our brain to be focused and present in our daily lives, what Dan Harris calls the "off the bench benefits" of meditation. The goal of meditation is not to empty our mind, which is impossible, but to focus on the present in spite the narrative in our head.

### **Basic Breath Meditation**

In my practice, when working with new student meditators, I recommend practicing 5 to 10 minutes a day. I also recommend finding a group or meditation coach in your area to help you grow and refine your practice. Set a timer so you are not worried about the time; 5 minutes is a very good and doable start.



**Read the instructions below**

***When comfortable -set your timer for 5 minutes and begin***

Find a comfortable position in which to sit for this period. As you allow your eyes to gently close, tune into your body and make any minor adjustments. It can be helpful to remember our intentions of both ease and awareness. Sit in a way that feels comfortable but alert.

*We'll start with a few minutes of concentration practice, just to help our minds settle and arrive in our present time meditation experience. Take a cleansing breath in and feel how the breath awakens your senses. As you breath out, imagine breathing out any tension, stress, or anxiety.*

*Now allow your body to resume its natural breathing and see where in the body you can feel the breath. It may be in the stomach or abdomen, where you can feel the rising and falling as your body breathes. It might be in the chest, where you may notice the expansion and contraction as your body inhales and exhales. Perhaps it's at the nostrils, where you can feel a slight tickle as the air comes in, and the subtle warmth as your body exhales.*

*You can pick one spot to stay with for this meditation practice. As you become a witness to your breathing, we will use "labeling the breath" as a technique to help you stay focused. As you breathe in with awareness say silently to yourself "in" and on exhaling, say silently to yourself "out". Remember that labeling the breath is a tool to help build concentration and focus and is not a measurement of how good a meditator you are.*

*You will notice your mind wandering. When your mind wanders, and it always will, we are being offered an opportunity to cultivate mindfulness and concentration. Each time we notice our mind wandering, we're strengthening our ability to recognize our experience. Each time we bring the mind back to the breath, we're strengthening our ability to focus on an object in the present moment. Treat this as an opportunity rather than a problem, and return to your "in" breath.*

### **Resources**

10% Happier: Dan Harris

Meditation for Fidgety Skeptics: Dan Harris, Jeff Warren, Carlyle Adler

Wherever You Go There You Are – Jon Kabat-Zin

Arriving at your own Door: 108 Lessons in Mindfulness – Jon Kabat-Zinn

No Time Like The Present- Jack Kornfield

### **Meditation Apps**

10% Happier

Calm

Simply Being

## PAF EVENT & FUNDRAISING SPOTLIGHT

### UPCOMING/ONGOING EVENTS

- 9/2018- 13th Annual Tailgate Party & Corn Hole Tourney for PAF, Gahanna, Ohio
- 10/20/2018– PA Family Day, Columbus area, OH (more info to follow)
- 10/21/2018- Team Propionic Acidemia Runners participate in the Nationwide Children's Hospital Columbus Marathon

### PAST EVENTS & CAMPAIGNS:

- 10/2017-Michelle's Crop, OH - \$2500
- 10/2017-Tara Gerlach Columbus 1/2 Marathon-\$550
- 1/2018 - Branch Superbowl Party, IN - \$570

### CORPORATE MATCHING GIFTS AND VOLUNTEER HOURS DONATIONS:

- **Corporate Matching Gifts:** This may enable you to double your donation. Check with Human Resources to see if your employer matches. It makes a big difference.
- **Volunteer Hours:** Some companies have a volunteers program and will donate based on your volunteer hours. PAF is always looking for volunteers. Please check with Human Resources to see if they have a program.

If you have anyone you would like to have us add to our campaign, please e-mail [paf@pafoundation.com](mailto:paf@pafoundation.com).

**FACEBOOK:** Thank you to all of our Facebook fundraisers. There were a record number of facebook fundraisers set up in the past 2 months for PAF Thank you for making a difference.

**INTERNET:** Thank you for using Igive, Goodsearch and AmazonSmile and designating Propionic Acidemia Foundation as your charity. Every dollar counts.

### DEDICATED GIFTS FROM INDIVIDUALS:

Among the many contributions received, the following is a list of some that were dedicated to those who have inspired the giver.

- **In Honor Of:** Ruby Dietz's 1st birthday, Carson Alfano, Kristin Boecker, Sean and Courtney Callahan, Brandon Napiwocki, Harvey Franks 80th Birthday, Lucy Harding, Dylan Jaenke, Nalani Johnson, Reuben Kleckley, Mr. and Mrs. Steven A. Lenert, Kate Lowry, Dania and Edgar Jr. Martinez, Leah Masten, Zach Matz, Connor McKillop, Michael J. Messersmith, Gabrielle Millett, Reuben Kleckley, Mel and Elise Russell, Laurel Sonntag's Birthday, Talli Smith, Maren Stecken, Isabella Velazquez, Chase Workman, Brett Young
- **In Memory Of:** Bob Buck, Alice and John Dawe, Sharon Esses, Alexa Faith Cardone, Kerrie Lynn Fessler, Jordan Franks, Vincent Franze, Nicholas Phillips, Dax G. Ross, Angelica Stageman

## OAK LAWN-HOMETOWN MIDDLE SCHOOL AND PAF

### A budding partnership



Students at Oak Lawn-Hometown Middle School are always looking for ways to make the world a better place, so it was no surprise when they heard about Propionic Acidemia they wanted to help.

Propionic Acidemia is an inherited disorder in which the body is unable to process certain parts of proteins and fats properly.

Social Studies Teacher at OLHMS, Sophia Georgelos, heard about a program through the Propionic Acidemia Foundation, from a fellow Teacher. The program provides birthday cards to children battling Propionic Acidemia from kids all over the world.

The entire 6th grade and even team 8a decided to take part in writing birthday cards for the "warriors" as they are called, but decided to do more and made holiday cards before winter

break. "We just wanted to show the kids what one small gesture can really do. Before the break, we wrote these letters during PBIS time, and some kids even came over lunch to do more. Now, we are partnering with this great foundation and bringing smiles to these kids fighting harder battles than we might ever face," said Georgelos. The foundation was overwhelmed with the generosity of the students and decided to partner with the middle school to make more birthday cards as part of the Warrior Birthday Club. Parents even wrote back to the students, thanking them for their generosity and sending a holiday card to their son who battles the disease.

Source: <https://d123.org/category/news/>



**We are so grateful to the students and staff at Oak Lawn-Hometown Middle School who graciously took over the Warriors Birthday Club!**

## ANNABELLE'S STORY (CONT. FROM PG. 3)



The highest her ammonia has gotten post-transplant has been 98 (from frequent vomiting), and her new normal on a "good day" now averages in the 30's. It's also been amazing to see the developmental progress she's

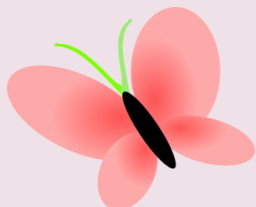
been making post-transplant. She's so much more alert, and her overall energy level and muscle tone have increased greatly.

Since Annabelle's transplant we have been able to come off of Carbaglu and Bicitra, but she is still receiving Carnitine (which we were told she'll be on for the rest of her life). Her feeding skills are still lacking, and getting her to eat by mouth is still a struggle. However, she's getting feeding therapy and making progress so we are hopeful that she'll eventually eat enough food by mouth to come off of her formula. Her protein intake can be less restricted now, but since she's primarily tube fed her metabolic dietitian has been conservative (1.3g/kg) and hasn't tried to push her protein as long as her amino acids stay within the normal

range. The main food advantage post-transplant is that we don't have to weigh her solid foods now and we just go by the nutrition label. Also, if she throws up we don't have to immediately pump more formula back in her; now we just let it go unless the vomiting becomes excessive. There's a lot more wiggle room in her overall stability, and we aren't "living on the edge" every single day like we were pre-transplant.

We know the decision to transplant your child is a difficult one, and we're very open to discussing our journey with any families that would like to speak with us. Please feel free to see more about our journey at [www.CotaforAnnabelleGM.com](http://www.CotaforAnnabelleGM.com) where you'll be able to find our blog posts, as well as, a link to Annabelle's Facebook Page where you can see photos and videos from our transplant journey.

Sincerely,  
Mike, Charity, and Annabelle



**Thank you for all donations and the kind notes we receive throughout the year. Your support overwhelms us and continues to be a source of inspiration. PAF couldn't do what we do without your incredible support.**

***We want to hear from you! Have a PA story to tell, event to promote or news? Fall newsletter submissions due by August 1, 2018!***

### ***Help Us Find the Cure!***

Name \_\_\_\_\_

Please send an acknowledgement to:

Address \_\_\_\_\_

Name \_\_\_\_\_

City, State, Zip \_\_\_\_\_

Address \_\_\_\_\_

Phone \_\_\_\_\_

City, State, Zip \_\_\_\_\_

E-mail \_\_\_\_\_

Enclosed is my contribution of \$\_\_\_\_\_ in honor of/in memory of \_\_\_\_\_

- By providing your e-mail address you are opting in to receiving e-mails from the Propionic Acidemia Foundation. We will not share your information with those outside of the foundation.
- If you work for a company that has a matching program, please include the matching form.
- Please mail your check made payable to: Propionic Acidemia Foundation 1963 McCraren, Highland Park, IL 60035

***Thank you for making a difference.***

SEARCHING FOR A CURE  
HOPE FOR OUR CHILDREN

Propionic Acidemia Foundation  
1963 McCraren Rd.  
Highland Park, IL. 60035



Phone: 1-877-720-2192 toll free  
Fax: 1-877-720-2192  
E-mail: [paf@pafoundation.com](mailto:paf@pafoundation.com)  
Website: [www.pafoundation.com](http://www.pafoundation.com)

## PROPIONIC ACIDEMIA FOUNDATION

### Board of Directors

Jill Chertow Franks, President  
Brittany Smith, Treasurer  
Angela Waits, Secretary  
Maria L. Cotrina  
Ann Marie Young

### Medical Advisory Board

Gerard T. Berry, M.D.  
Barbra Burton, M.D.  
William Nyhan, M.D., PhD  
Mendel Tuchman, M.D.

### Newsletter Committee

Julie Howard  
Jennifer Mouat



Join and follow  
Propionic Acidemia  
Foundation on Facebook

**PAF volunteers and  
board members are  
needed!**

[paf@pafoundation.com](mailto:paf@pafoundation.com)

*Publication Note: The PAF Newsletter is published twice a year. Readers may subscribe by writing to PAF, registering online or calling 877-720-2192. Letters and article submissions are welcome for consideration and may be sent to [paf@pafoundation.com](mailto:paf@pafoundation.com) or mailed to Propionic Acidemia Foundation 1963 McCraren, Highland Park, IL 60035. If you would like to be removed from our mailing list or receive the newsletter via email, please contact us.*