

Propionic Acidemia Foundation

VOLUME 1, ISSUE 3

FALL 2007

FOUNDATION NEWS

The Propionic Acidemia Foundation is pleased to announce our new collaboration with Coriell Institute of Research to accelerate knowledge on Propionic Acidemia. You can read more about this exciting project on Page 2.

We would like to thank all of you for making this a record fundraising year and record year for grant disbursements. All of this could not have been achieved without your help. Thank you. This is just the beginning. We are hopeful that PAF will be able to continue to fund researchers devoted to finding better treatments and a cure for PA and know that our dream will become reality with your help.

FINANCIAL REPORT - FISCAL YEAR 2006-07

Revenue:		
Contributions		\$84,397.99
Interest Income		\$ 3,118.51
Total revenue		\$87,516.50
Expenses:		
Research Grants awarded	\$80,989.04	
Programs & Outreach	\$ 931.35	
Fundraising	\$ 229.80	
Operational Expenses	\$ 307.36	
Conference Expenses	\$ (76.57)	
Total Expenses	\$82,380.98	
Cash Assets August 1 st 2006	\$54,803.25	
Cash Assets July 31 st 2007	\$59,938.77	

Board disclosure

Jill Chertow Franks is the sister-in-law of Michael Rosenson.

Me Volunteer?

Of course you!

The Propionic Acidemia Foundation needs people like you to write articles, review brochures, participate in surveys, help with programs and fundraising. Whether you want to volunteer on a regular basis or just occasionally, there are a number of volunteer opportunities available to you. Any time you can give will be greatly appreciated and you'll feel great by helping to improve the lives of all individuals affected by Propionic Acidemia.

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PAF AND CORIELL ANNOUNCE COLLABORATIVE PROJECT

The first samples are being processed and will hopefully be available to the scientific community in early 2008.

Propionic Acidemia Foundation and Coriell Institute for Medical Research in Camden, NJ have initiated a collaboration to provide the NIGMS Human Genetic Cell Repository with samples from subjects and their families with PA and a thorough set of clinical, molecular and laboratory data. We have developed an extensive questionnaire with the expertise of the Medical Advisory Board of PAF which will be administered for each submission.

Coriell Institute for Medical Research has pioneered Biorepositories for the study of human diseases. Most of the repositories at Coriell are sponsored by agencies of the NIH. The samples are used by scientists for gene discovery, functional assays, reference materials in assay development and genomics. The goal of providing the samples is to improve the understanding of human genetics and human diseases.

The Human Genetic Cell Repository, which is sponsored by the National Institute of General Medical Sciences (NIGMS), has cell lines and DNA samples from more than 500 inherited diseases. These cell lines have been an important resource for the identification of genes in which a mutation or chromosomal defect leads to the disorder. Yet, this is often only the first step. For some inherited disorders, it has been possible to perform a "genotype-phenotype" analysis, connecting the patient outcome with the type of symptoms displayed by the patient. For others this connection is not obvious. One possibility is that the outcome of the disease is due to variants in other genes. In order to investigate these possibilities it is necessary to have excellent longitudinal clinical information in addition to the laboratory analyses. To build these resources we have begun establishing interactions with groups which interact directly with the patients on a continuing basis.

As noted above it is critically important, not only to find the gene which is the primary cause of the disorder, but also to understand the genetics and other factors which could lead to an understanding of the way the disease unfolds.



Cells at Coriell are stored in liquid nitrogen at -320°F (-196°C). This extreme cold permits cells to be revived for more than 40 years.

To achieve this Coriell and PAF have developed a protocol whereby subjects who submit samples to the Repository for the preparation of a cell line, will also be recontacted in the future so that the course of the disease can be known to the scientists who obtain the cell lines or DNA from the Human Genetic Cell Repository.

To protect the identity of the subjects and their families, Coriell developed a protocol, approved by the Institutional Review Board, where the Propionic Acidemia Foundation would be the only ones who would be able to connect the identity of the subject with the sample in the Repository.

PAF will also follow up on the patients every few years to create a database on the status of the patients in addition to information about cells derived from the patients. Our goal is to create a new type of resource for the study of rare diseases. At the time of this newsletter we have received our first samples from this collaboration and they are currently being processed. We hope to have them available to the scientific community in the spring of 2008.

If you are interested in finding out more information or would like to be mailed a kit, please contact Jill Franks at paf@pafoundation.com or 1-877-720-219.

FUNDRAISING

This past year was the most successful fundraising year we've had; we raised over \$87,000! With these funds we were able to fund three research projects. Several affected families held their own fundraising events, which greatly impacted the projects we could fund. However, fundraising efforts do not have to encompass large events to make an impact on research and the lives of those with Propionic Acidemia. Effective fundraising can be done by very simple acts. Here are some examples:

- Make a donation to PAF yourself.
- Make a donation in honor of a loved one or an event.
- Ask your employer about matching donations to non-profits. If they match, fill out the appropriate forms after making your donation.
- Set Goodsearch.com as your homepage on your computer and use it as your search engine. Goodsearch has a new shopping site called Goodshop, there is nothing to sign-up for, just link through the site and a portion of your purchase price will be donated to PAF.
- Ask 10 friends for a \$10 donation. You have quickly raised \$100!
- Put donation request cards in your holiday cards. Contact Brittany at paf@pafoundation.com or 877-720-2192 to have donation cards sent to you.
- At the death of a loved one, request donations to PAF in lieu of flowers.
- Turn in used brand-named ink cartridges to Office Max or Staples and send your gift certificate into PAF, then more funds can be used for programs and research.

In the past 1 1/2 years, PA Families and Friends have used www.goodsearch.com over 24,000 times to raise over \$240.00 for our cause. Keep searching and telling everyone you know about Goodsearch and Propionic Acidemia Foundation.

How to set Goodsearch.com as your homepage

Internet Explorer

Right click on Internet Explorer

Click on 'Properties'

On 'Home page' enter 'www.goodsearch.com'

Click apply

Go to www.goodsearch.com

Under 'Who do you goodsearch for?' enter 'Propionic'

Click 'Verify'

Now search using Goodsearch and raise \$.01 for PAF with each search

Now shop with Goodshop and have a percentage of your purchase price donated to PAF.

For more information, email PAF at paf@pafoundation.com.

In kind donations

Turn in your brand named ink cartridges to Staples or Office Max and send PAF your gift certificate

Postage Stamps

HP 56 ink cartridges

Thank you cards

Samsung ML-2510 Toner

Mailing labels 2"x4"



Raised over \$1068.00 with iGive

PA FAMILY SPOTLIGHT - AIDAN PRAGASAM

Birth of an angel - Miracles still happen and my son, Aidan Pragasam, is a living miracle by me. Aidan turned 13 this year. He is a happy, cheerful, handsome boy who was diagnosed with Propionic Acidemia when he was 3 weeks old. Aidan had a difficult birth that was further complicated with a Steph infection. He was released after spending two weeks in IICU and everything seemed normal at that time. A few days later, we noticed that he seemed lethargic and inattentive with no inclination to feed. Repeated consultation with his pediatrician went nowhere and we were getting increasingly concerned that Aidan was fast approaching a comatose state. An unexpected call from the University of Pittsburgh that had screened Aidan's blood for PKU revealed very high content and we were asked to rush Aidan immediately to the Children's Hospital in Philadelphia. Aidan was seen immediately by a team of experts led by Dr. Paige Kaplan and admitted into the IICU (again) and placed on IV. He had very high levels of ammonia in his system. After several blood /platelet transfusions and a stay in the IICU for three weeks, Aidan was moved to the general pediatric ward where he stayed for a few more weeks before being allowed to go home. During his first year of life, Aidan was repeatedly hospitalized and each time it was a traumatic experience for all of us. He had spent over seven months in the hospital during the first twelve months of his life. The second year was slightly better with hospital stays that lasted days instead of months.



Selvi, Aidan (13) and Brenden (9)

Tough beginnings - During the first few years the going was tough on all of us. It was extremely difficult for Aidan as we observed several global delays in him and he was late in hitting every developmental milestone. He did not walk until he was three years old and was not verbal until he was six years old. Aidan had a regiment of therapies as he was growing up and even though the therapies have dwindled over the years, he still continues to receive therapy on a regular basis.

To put our ordeal into perspective, I have to give a little background on our life before Aidan. My husband Ravi migrated to the US from India for his graduate studies and I followed him here after our marriage. Aidan was born on March 23, 1994 exactly a year after I came to the US. The culture shock, the extreme cold weather and the loneliness of leaving my family and loved ones behind, just added to the complexity of the situation. Our life together was an emotional roller coaster and we had no family or real friends who could offer support at these difficult times. We could not partake in social gatherings since we always ended up leaving early due to Aidan's needs and as time went by, people started leaving us alone since they did not have the emotional maturity to understand our situation and offer support or they did not want to be bothered. We had no one to turn to except each other and this brought us closer to each other since we were committed to giving our son the best life possible and I found the purpose of my life revealed. For me, Aidan was never just a child, he is an angel sent from heaven on a mission. I am not saying this because I am his mother, but because everyone who comes in contact with him says the same. There have been strangers who have walked up to me and said your child's face is angelic. His cheerful childish ways and endurance to pain in the worst of situations and his pure love for people is a miracle by its own.

Better times - Aidan was 3 years old when we moved to San Jose California. After a year of difficulty finding a good team of doctors, we became members of Kaiser Permanente with an excellent team. His nutritionist Elaina Jurecki has been the lead in shaping Aidan into what he is today. Her insights, knowledge and expertise have been a godsend to us.

We became very active in a South Asian organization called Jeena, a support organization for special need kids consisting of families with kids like us who were also looking for a social network and who had faced the same level of ostracization from their social peers. The challenges that we faced as a family in the social front led me to play an active part in establishing this organization along with its founder. In this process we have met many good people and made many good friends. We had found our bearings and with the passing of years we have formed stable friendships that could last a lifetime.

Aidan has been attending a special education school in San Jose with the same Elementary school teacher/classroom for almost 5 years. Aidan and his teacher grew very attached to each other. When he graduated from elementary school she sent him off to middle school with a brand new Mac computer, a collection of software that can last a lifetime and books on several sub-

MIRACLES STILL HAPPEN - THE MIRACLE CONTINUES

jects that would serve a whole classroom full of kids. He is settling down slowly in his new school and adjusting to his new teacher. His new friends have taken to him well and are very fond of him. Besides school, he goes to Score Computer Based learning center. He is very computer savvy and knows his way around with PC games and software. He loves web surfing and his current favorite is Youtube where he watches his favorite TV theme songs such as Barney, Teletubbies, Scooby-Doo, Jeopardy, Wheel Of Fortune over and over again. He also goes swimming and attends gym classes in Jeena.

Aidan has a younger brother Brenden who is 9 years old and who adores him. Aidan loves having his brother around. The beginning challenges we had in devoting equal amount of attention to both kids got easier as years went by. We have now learned to balance our time and life between the two kids and also our careers. We have a great nanny who helps us in taking care of the kids. We are able to travel places and do things that normal children do, like visiting theme parks, going to shows, going for bike rides etc. Aidan is finally able to ride a specially made bike with three wheels and pedals designed for children with physical difficulties. He wears braces to provide ankle support and uses a wheel chair when we go on long walks or have to travel to unknown places. He is very interested in music and dancing, especially Indian music. He has an incredible sense of rhythm and it amazes me when I see him move to the beat even if he cannot use his legs very well. I often catch him dancing to music in front of the mirror. He has also participated in dances and performs in Jeena's annual day celebrations.

Successful completion of a decade - He continues to be a miracle since his doctors gave us a very discouraging prognosis about his survival during the initial years of his life. So when he turned 11, I decided to publish a website for him. His website has brought in many contacts internationally and helps me keep in contact with people who have children with the same disorder in the far corners of the world, where they are not blessed with the quality treatment we get here.

Working towards a cure – the Fundraiser - I am a professional classical dancer and coupled with Aidan's interest in Indian music and dance along with my husband's and parents support, I decided to start an Indian dance school. The dance school has grown today quite a bit. Aidan completed 13 years this year and that made me believe that something needed to be done to raise awareness of this very rare disorder that even sounds alien to most pediatricians and medical professionals. Awareness is essential to raise funds and a CURE is needed to help our children. And it has to happen right now so that we can see some benefit from it during our lifetime. To keep the research going, we need funds. Being involved in a non-profit organization made me realize how hard it is to get a grant approved or to get donations coming in. So this year I decided to conduct a fundraiser for PA Foundation keeping in mind all my goals in increasing awareness and raising funds. My family supported me in this effort especially my kids and my husband.

Along with the students and parents of my dance school we put together a dance performance and a fundraiser on May 12th 2007. My friends helped me by putting up a silent auction and volunteering their time and energy. The event was a huge success and was attended by almost 500 people. Elaina Jurecki, Aidan's nutritionist gave a presentation for about 20 minutes on the topic of PA and why funds were necessary to conduct research looking for a cure for this rare and devastating disorder. To many of the audience it was a revelation that such a disorder existed. There was no admission ticket charge and all the funds raised through donations and the silent auction were donated to PAF for research. People not only donated during the event, but also several days after the event had taken place. Funds close to \$3500 were raised during the event. In addition to the fundraising, there was quite a bit of media coverage on the event and its cause. I believe that this is just the beginning. With my angel at my side, I am hoping to continue to contribute in whatever small way possible to help support this miracle and keep it alive. I have faith and belief that before long we will see the day when all our children will be CURED. Yes, the miracle still lives and will continue to thrive!!

Selvi Pragasam, mom to Aidan and Brenden



Dancing for a Cure

PA FAMILY DAY AUGUST 11TH 2007

Dear Family and Friends,

Lucy and I attended the second annual PA Family Day in Chicago on August 11. Jill Franks and her family graciously opened their home to host the event again this year. Fortunately Jill and Steve live in a mansion because that is just what we needed for the crowd of about 40. There were seven children and one adult with PA in attendance along with many parents, grandparents and siblings. This is double the numbers from last year. Yeah!!!



PA Family Day Group Photo

The day was packed with chatter as we all enjoyed meeting new friends and reconnecting with old ones. It was such a fun, relaxed atmosphere, sort of a family reunion as many of us met people we had heard about on the web site or talked to on the phone. What fun to put faces with names and be able to pick other brains for new ideas. There were kids running around everywhere. The basement playroom was one of Lucy's favorite places because of the swing and the dollhouse, however I don't think Ryan was too happy about the little kids messing up his Hot Wheels cars. Ha!



Lucy loves her spaghetti.

Lucy loved meeting all the other kids. "They have Propionic Acidemia just like me Mommy!" "Look Mom, they have a tube feeding just like me."

Her highlight was showing off by eating spaghetti and drinking juice out of a wine glass for everyone. We especially enjoyed meeting the young members of the group, the Ohio girls – Allison and Gwen.

Thanks to many local retailers, formula companies, and low protein food companies for helping out with the great food. We had Panera bagels, fruit and veggie trays from Sunset Foods, and the most amazing Chicago style

pizza from Lou Malnati's. The low protein choices were amazing – cookies, cheese, crackers, muffins, cereal, pasta, pretzels, candy, chips and more from Applied Nutrition, Cambrooke, and Ener-g. For sure no one went away hungry.

Brittany shared Talli's life book and I took my scrapbooks.

PA FAMILY DAY

It was fun to share life experiences and activities with everyone. Jill presented us all with a new opportunity to participate in research for the cause. A DNA repository is being collected by Coriell Institute for Medical Research. Jordan's physician and member of PAF Medical Advisory Board, Dr. Barbara Burton, was on hand to draw blood from all of us who wanted to participate that day. Take home kits home to have blood drawn at a later date were available (and still are if you haven't contributed yet). This was a great opportunity for those of us affected by PA to provide scientists with genetic cells for future study.

The great weather gave us the opportunity to enjoy the outside. Jill's backyard includes a huge play set, nice shady area, a bubble maker, and a sprinkler. Even the big kids got into the games. Lucy really liked playing in the sprinkler with Gwen, who was soaked and loving every minute in the wa-wa. Many smiles were shared.

I am so happy we were able to make the journey to join in for PA family day. It was a great day. The time with those who truly understand our experiences and struggles is so limited, I will value this opportunity always. We are not alone. In fact, next year I am thinking of hosting the day in Kentucky. Any suggestions? Reach me on the PAF list serve.

Love and Good Health to All – Julie Harding (Lucy's mom, age 9, PA)

A special thanks to the families that attended: The Ellis Family, The Franks Family, The Gordon Family, The Harding Family, The McKinley Family, The Mouat Family, The Napiwocki Family and The Smith Family

Help Us Find the Cure!

Name _____ Please send an acknowledgement to _____
Address _____ Name _____
City _____ State _____ Address _____
Zip code _____ City, State, Zip _____
Phone _____
E-mail _____

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

If you work for a company that has a matching program, please include the matching form.

Please send your check made payable to: Propionic Acidemia Foundation

1963 McCraren Rd.

Highland Park, IL 60035

Thank you for making a difference.

SEARCHING FOR A CURE
HOPE FOR OUR CHILDREN

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We would like to thank our Medical Advisory board for all of their support, input, advice, and knowledge. We could not do it without you.

We would like to acknowledge and thank each of you personally for making a difference for families affected by Propionic Acidemia.

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