SEARCHING FOR A CURE HOPE FOR OUR CHILDREN

Propionic Acidemia Foundation

VOLUME 1, ISSUE 5 FALL 2008

Special Double Issue!

PA Family Day is a success

This year's event was held at the beautiful Harding Home in Lexington, Kentucky. We were thrilled to have over 55 people in attendance including 12 families affected by PA from Georgia, Illinois, Indiana, Kentucky, Missouri, Ohio and Washington.

We were joined by Dr. Charleton Mabry, endocrinologist at UK hospital, who spoke about the first documented case of PA, pediatrician Dr. John Riley and Carla Barrowman, who spoke about case management, futures planning and supports for community living. PAF held a board meeting which gave all of the families a chance to participate and hear the latest information on PAF. The children had a blast playing outside and PA Family Day even made the Lexington evening news.

Photos are online at www.pafoundation.com We hope you will join us next year!

Our children with PA included: Gwen 28 months, Brandon 9 (brother Matt 12), Amber 12, Tiffany 8, Kaitlin 9 (sister Maddy 7), Talli 6 (brother Max, 12 months) Allison 5 months, Hogan 8, Trent 6, Lucy 10, Jordan 8 (brother Ryan, 11) (not pictured Chase 10)

FUNDRAISING

Thanks to your support we had our best fundraising year to date. Thank you!!!

See page 10 for fundraiser updates.

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ANNUAL REPORT: FISCAL YEAR 2007– 2008

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.

FINANCIAL REPORT

Revenue:	
Contributions	$92,269
Interest Income	$ 2,102
Total revenue	$94,371
Expenses:	
Research Grants /Exp.	$82,404
Programs & Outreach	$ 3,605
Fundraising	$ 233
Operational Expenses	$ 1,196
Total Expenses	$87,438
Cash Assets 8/01/08	$66,872
Cash Assets 7/31/07	$59,939

BOARD OF DIRECTORS/OFFICERS

Jill Chertow Franks, President

Brittany Smith, Secretary, Treasurer

Janice S. Boecker

Michelle Burns

Jennifer Mouat

Michael Rosenson

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

PROGRAM ACCOMPLISHMENTS

RESEARCH: 16 samples from affected individuals and 23 from carriers have been submitted to the DNA Repository at Coriell Institute for Medical Research in Camden, NJ. For more information or to participate, please call PAF at 1-877-720-2192 or email research@pafoundation.com

Grant Disbursements:

$30,000 - Jan P. Kraus, Ph.D., University of Colorado at Denver and Health Sciences Center (UCDHSC) Denver, CO: "Crystallization and structure determination of human propionyl-CoA carboxylase"

$40,000 - Jan P. Kraus, Ph.D., UCDHSC Denver, CO: "Genotype –phenotype correlations in PA"

$12,500 - Dr. Michael Barry, Ph.D., Mayo Clinic, Rochester, MN: "Gene Therapy for Propionyl CoA Carboxylase Deficiency"

OUTREACH:

Conferences: PAF attended and exhibited at 2 conferences, Society for Inherited Metabolic Disorders and Genetic Metabolic Dietitians International, at which we were able to network with metabolic doctors, dietitians and researchers and share information about PAF.

PA Family Day: See cover story.

HOW CAN I HELP?

Propionic Acidemia Foundation is run 100% by volunteers and we couldn't do it without you.

Ways you can help:

- Search or play games to earn tickets which can be donated with SearchandGive.com
- Search the internet through GoodSearch.com or shop through GoodBuy.com
- Search or shop through iGive.com
- Sign up for eScrip
- Sell items on Ebay using MissionFish. You can pick what percentage you want to go to PAF.
- Turn in used ink cartridges to Staples and give them the PAF phone number (1-877-720-2192) to earn gift certificates for office supplies.
- Make a donation.
- Plan a fundraiser.
- Volunteer your time and talents.

GoodSearch
Raised over $440.54

Raised over $1564.00

MissionFish
Raised over $314.97

eScrip

FAMILY SPOTLIGHT - KIRSTYN TRIPP

After ten years of infertility issues, Kirstyn was on her way! She turned 4 on Saturday Aug 17 and mom said her birthday was bittersweet as it is every year. Like so many PA families, her parents were told in the first week that their daughter would likely not live through her initial crisis. When Kirstyn was born she had ABO incompatibility which is basically severe jaundice, so she was transported to Shands NICU to get her liver to kick in. The first 7 days there Kirstyn became very lethargic and stopped eating. One of the nurses even scolded mom saying she couldn't take her child home until she proved she could feed her. The next day Kirstyn crashed, falling into a coma. Ammonia was over 700 and she spent 9 weeks in that NICU, during which time she had a core team of nurses assigned to her because of her rare condition and bleak prognosis.

After that she went the entire first year without further need for hospitalization, but has had 4 since then. She's had three blood transfusions of packed red cells. She was diagnosed with mild cardiomyopathy at 18 months, so she has an EKG and EEG every 6 months. Stomach bugs are the hardest thing, causing 2 hospitalizations due to excessive vomiting, ketones and high ammonia. In December 2005 she spent 2 days in the hospital for a terrible ear infection that was first misdiagnosed as seizures because she was vomiting then zoning out. In March 2006 she was in overnight for a respiratory infection and high ammonia. Her mom said "Kirstyn gets sick really quickly! Once she reaches moderate ketones, she will need to go straight to the hospital or her body will begin rapid decompensation."

Kirstyn had OT & PT her first 3 years through an Early Intervention program. At the age of 3 she broke her collar bone in school because she didn't reach to catch herself during a fall due to her low muscle tone. She's clumsy when she walks, but has always been very active. Her mom said "If she gets red faced, we have to give her extra calories or she will spill ketones within an hour of hard play. We learned that the hard way during her 3rd birthday party at the park."

Last year, at age 3, test results showed Kirstyn had reading and comprehension skills of a 6 year old and counting and number abilities of a 5 year old. She's going into her 2nd year of pre-K geared toward medically impaired children with no cognitive delays. She's quite bold and independent and it's very difficult to keep her on a task she doesn't want to do. She loves to sing, draw, and paint. She has very low muscle tone in her hands so she really resists anything that involves use of hands, like using scissors. Due to her low muscle tone Kirstyn still receives PT through school. She has a lot of behavioral issues and doesn't like to listen. Her school has recently considered testing her for ADHD, even though they're also considering testing her for the gifted program.

She's just started her first activity, tap dancing & loves it! Recently Kirstyn learned to ride a bicycle with training wheels, which she

got for her birthday. Playing on the computer is one of her favorite pastimes. She doesn't like her little brother, Cason (20 months), and often asks her mom to "Please send him back to the Little Brother Store." She picks out all of her own clothes and doesn't care what people think. ☺ And, she loves to do housework.

Kirstyn eats 100% by mouth except when she's sick. With a packed lunch from her mom she eats so much better to at school with friends around. Her mom said "We always know when she's getting sick because her first symptoms are always spilling ketones and she stops eating. For this reason she has a g-tube". Like many young children she has a weird diet consisting largely of french fries, pancakes, hash browns and loves to eat at McDonalds and Bob Evans! Her mom said she likes ketchup on mini frozen waffles and calls them mini pizzas. She would eat ketchup on everything (anything mom lets her put it on!) if she could. She doesn't care much for sweets, only an occasional M&M. At parties she'll tend to lick a bit of icing to feel like part of the group.

As far as potential complications from PA, she's doing amazingly well. As of this year her doctors said she only needs labs 4 times per year! Her restricted amino acids typically run around the low end of normal and she's given a Valine supplement because it has always run below normal. Her Glycine is typically around 800.

Marsha, Kirstyn's mom, expressed her sincere appreciation for the support she's received from the other PA families through PAF saying, "We've been blessed to be part of this support community, which I've turned to for help on many occasions." She said they've even met a few other PA families because they live so close to Walt Disney World, so look her up if you're planning a trip!

Written by J Mouat; jenmouat@gmail.com
on behalf of Marsha, Ronnie, Kirstyn, & Cason Tripp, Florida
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AN OVERVIEW OF NUTRITIONAL MANAGEMENT OF PA

SPECIAL NUTRITION INSERT: KEEP THIS ISSUE AT HAND FOR REFERENCE.

Elaina Jurecki, MS, RD

General Overview of the Metabolic Pathway Dysfunction

Much has been learned about propionic acidemia (PA) over the past several years. We know that proteins are broken down into amino acids in the body which can then be utilized to make all the necessary body parts, such as skin cells, heart muscle, and liver tissue. The excess amino acids are further processed to provide energy for the body. An important enzyme, propionyl-CoA carboxylase (PCC), is needed to convert four specific amino acids, isoleucine, valine, threonine, and methionine, along with other nutrients such as odd chain fats, into important compounds which can then be processed to make energy. The PCC enzyme in individuals with PA is not functioning appropriately, leading to a build up of several acids, with the predominant one being propionic acid. These acids are called organic acids since they have been produced by the body, and hence, PA is termed an Organic Acidemia, or disorder leading to the excessive production of organic acids. PCC impairment results in the accumulation in propionic acid, 3-hydroxy-propionic acid, methylcitric acid, along with lactic acid and ketones. The accumulation of these acids impair the Kreb cycle, which is the primary energy producing cycle in the body. The Kreb cycle takes place in the mitochondria of the cell, which is considered the energy production center. Individuals with PA have shown compromised ability to make energy in their mitochondria. Not only can the acids that accumulate in PA interfere with producing energy, but they can also interfere with the body's ability to detoxify the waste products produced from amino acids leading to the accumulation of ammonia. Individuals with PA will present with frequent vomiting, lethargy, refusal to eat, and poor muscle tone resulting from the accumulation of organic acids, interference in energy metabolism, and a build up of ammonia in the body. A metabolic crisis can occur because of an inadequate calorie and/or protein intake, excessive protein ingestion, body muscle protein breakdown, and from other causes that lead to increased stressed states such as illnesses.

Sources of Propionic Acid

Studies have been conducted to determine what the primary sources for production of propionic acid were in PA. The investigators assessed 3 PA children and found that 50% of the PA production came from the break down of amino acids, 25% was produced by the natural bacteria found in the intestines, and the remaining 25% was assumed to be produced from the break down of fats used to make energy during fasting. The main type of fats that lead to the production of PA are called odd-chain fats which are made up of an odd number of carbon units in their chemical structure. Odd-chain fats are processed by PCC in order to generate energy. The results of these investigations suggested that the sources of propionic acid, other than from the four amino acids received from the diet and produced from muscle protein catabolism, are important sources of propionic acid and explain the need to implement additional treatment measures besides a dietary protein restriction. (Thompson, Metabolism, 1990)

Goals of Therapy

Goals of therapy are to promote normal growth and intellectual development by providing an adequate nutrient and fluid intake to promote anabolism (growth) while minimizing the accumulation of toxic chemicals in the body. There are many components to treatment in addition to dietary management including ensuring adequate physical activity, receiving the necessary therapies such as physical or occupational therapy, maintaining dental health, correction of anemia, and taking the appropriate medications, such as carnitine.

Dietary Management

Dietary management will vary widely from patient to patient depending on degree of the PCC enzyme function that each individual has and whether or not their condition presented very early when they were an infant versus later in life. It is important to tailor treatment to meet each individual's needs as one size does not fit all. Some individuals will be able to tolerate a fair amount of protein in their diet, while others will require a much more severe restriction.

Conventional dietary management of PA is aimed at limiting the flux through the protein/fat degradation pathway where the malfunctioning PCC enzyme is located, by restricting the amino acids, threonine, methionine, isoleucine, and valine, and with avoidance of fasting to decrease body protein and fat breakdown. These measures are important to help maintain metabolic stability, but the diet also has to contain an adequate amount of calories, protein, and vitamins and minerals to ensure sufficient nutrients are received to allow for growth and development. Additional treatment measures include supplementation of carnitine to correct for deficiencies of this important nutrient. Carnitine binds to many of the toxic metabolites or organic acids that build up in the body, detoxifying them and allowing for them to be excreted in the

NUTRITION MANAGEMENT CONTINUED

urine. Prevention and/or aggressive treatment of infections is also important to prevent the body from producing the toxic acids due to breakdown of body proteins and fats needed to fight the infection.

What does Carnitine do? It "binds to many of the toxic metabolites or organic acids that build up in the body, detoxifying them and allowing for them to be excreted in the urine."

In one study, PA patients were monitored after being fed and compared to after an overnight fast. The results showed an increased amount of toxic metabolites excreted in the urine in the fasting state as compared to the fed state. During the fasting state, the body breaks down fat stores to be utilized for an energy source. The investigators thought that during this process there will be the release of odd chain fats which cannot be properly processed due to the improper functioning of the PCC enzyme. They concluded that extended periods of fasting should be avoided in children with PA. (Thompson, Pediatri Res, 1990).

There have been several studies conducted to determine energy needs, or how many calories individuals with PA should have per day. In one study, conducted in the United Kingdom, they found a reduced rate of calories expended in PA patients, 80% of that predicted based on calculations for unaffected individuals. In contrast, a report from the Netherlands showed that the calorie expenditure rate was not decreased, but slightly elevated. These investigators saw a strong correlation observed between their calorie expenditure rate and the amount of special formula consumed. This group concluded that individuals with PA do not have a decreased rate of calories used by the body, and that use of adequate amounts of medical formula was important to establish a normal rate of calorie expenditure. (Feillet, J Peds, 2000) (Van Hagen, JIMD, 2004). Obviously more research is needed to help understand how many calories are needed for individuals with PA. This is really important as offering too many calories will lead to an excessive rate of weight gain which can compromise the individual's ability to move around. Providing an inadequate amount of calories can lead to body breakdown of protein and fats to produce energy leading to an excessive production of toxic acids and metabolic instability. For now, the best approach is to work closely with the metabolic clinic to make sure a sufficient amount of calories are being provided to ensure an appropriate rate of growth and weight gain.

Metabolic Formulas

There are many metabolic formulas available for all ages and made up of different compositions. In general, these formulas provide the majority of the protein intake, from a source that limits or excludes the offending amino acids, isoleucine, valine, threonine, and methionine, but is rich in the other amino acids. These formulas will also provide an important source of calories, vitamins, and minerals. The variety of formulas has significantly increased over the years. There are formulas specifically tailored to meet the needs of infants including Analog XMTVI from Nutricia, Propimex1 from Abbott Labs, and OA1 from Mead Johnson. There are also several formulas designed for older children and adults including Maxamaid and Maxamum XMTVI from Nutricia, Propimex2 from Abbott Labs, and OA2 from Mead Johnson. These formulas are typically more concentrated in protein and not as calorically dense as compared to the infant formulas.

There are additional formulas including, OS2 from Milupa and MMA/PA Express and Gel from Vitaflo that are very concentrated in protein which can be used to treat older individuals. Some of the formulas contain carnitine and others will have small amounts of the amino acids, isoleucine and threonine. It is important to work closely with the metabolic dietitian to determine which is the best formula to use. Sometimes individuals with PA need more calories than what is provided in the formula, or can only tolerate calories without protein during illnesses. That is when a formula that only supplements calories, and possibly vitamins and minerals, come in hand. There are several of these types of formulas including Pro-Phree from Abbott Labs, PFD2 from Mead Johnson, and Duocal from Nutricia.

A study showed improved growth in PA patients when their intakes were adequately supplemented with a metabolic formula. These individuals had their intakes optimized to promote catch up growth by increasing the calories and protein provided by the formula by 25%. Results from the study showed improvement in the growth curves for weight, increasing from 26 to 49%, length increasing from 25 to 33%, and head size increasing from 43 to 54%. The investigators also saw improvement in certain nutrient levels in their blood, which were able to reach the normal reference ranges. They concluded that by providing energy and protein for patients at recommended intakes can promote catch up growth and improve overall growth and nutritional status. (Yannicelli, Molec Genet Metab, 2003).

NUTRITIONAL MANAGEMENT CONTINUED

There was one study that looked at how compliant PA patients were with following their diets. The investigators in this study looked at food records in terms of compliance and protein intake and found that 57% of the group over restricted their diet, taking in less natural proteins (those proteins found in foods, not from the metabolic formulas) than what was prescribed, versus 28% who liberalized their intakes taking in more than what was prescribed. Only 14% took in the equivalent amount of dietary protein prescribed. The investigators thought that the over restriction of protein was most likely due to a concern for compromising metabolic control or due to a failure to appropriately adjust the diet for growth. It is important to closely work with the metabolic clinic to make sure the diet is adequately meeting the PA patient's nutrient needs. (Scholl-Burgi, JIMD, 2004).

"...more research is needed to help understand how many calories are needed for individuals with PA… providing an inadequate amount of calories can lead to an excessive production of toxic acids and metabolic instability.

Anorexia, or loss of appetite, and vomiting are universal problems seen in children with PA, particularly in the first year and early in the second year of life. The reason for these symptoms is unknown, although investigators have found that these children have unprovoked gagging during episodes of metabolic crisis. It is possible that abnormal accumulation of organic acids can affect the central regulation in the brain of the gag reflex and appetite. The placement of a gastrostomy tube greatly facilitates routine feeding and home management of minor illnesses associated with increased ketone production and times of increased caloric requirements. Some individuals with PA have oral motor problems which can interfere with their ability to drink, chew, and swallow. Gastrostomy tubes can greatly facilitate ensuring that these individuals receive an adequate nutrient intake. They also greatly simplify the administration of medications. Consideration for placement of a feeding tube should be made prior to the child becoming so anorexic that it has impacted growth and development. As the child gets older, and more metabolically stable, they may no longer need a feeding tube, which can than be removed at that time.

Supplement with Essential Fats

Some dietitians will recommend a fat restriction to avoid potential sources of odd chain fats or due to lower caloric needs associated with compromised mobility. Under these circumstances, it is important to ensure that these individuals receive adequate intakes of linolenic and linoleic acids, which are fats considered essential and are needed to be consumed in the diet. Various fat levels were analyzed in 22 patients that had a variety of metabolic disorders in the metabolism of organic acids, including PA. The investigators found that these patients, which required protein restricted diets for treatment of their metabolic disorders, had significantly lower levels of eicosapentanoic acid (EPA) and docosahexanoic acid (DHA). These are fats that are synthesized by the body from the essential fats, linoleic and linolenic acids. The investigators concluded that their diets were most likely low in essential fats since the primary dietary sources of these nutrients are typically found in higher protein foods which are excluded from the diet. (Netting, JIMD, 2002). Some patients have been found to have higher levels of triglyceride fats in their blood, due to an increased release of these fats from the liver. Other authors report that synthesis of essential fatty acids is not affected in clinically stable children with PA, so further studies are warranted in this area. Dietary supplementation of DHA in patients with methylmalonic acidemia (MMA), a metabolic disorder which is very similar to PA, did not have any adverse effects during the study and they were able to normalize their triglyceride levels (Aldamiz-Echevarria, JIMD, 2006).

The research in the area of fat metabolism is relatively new, and there is much to be learned to determine what the optimal amounts and types of fats that should be included in the diets for individuals with PA.

Monitoring Metabolic Control

There are several methods that have been reported in the scientific literature on how to monitor metabolic stability. Some clinics will monitor the urinary levels of metabolites, including urea, which reflects protein catabolism, along with measuring the amounts of organic acids, such as methycitrate and 3-hydroxy-propionate, that have accumulated. Measurement of urinary ketones at home can be done on a fairly regular basis to help assess metabolic stability. Propionic acid can be measured in the blood and may represent a more accurate measurement of metabolic control, but this measurement is only done by a few labs in the country. Odd chain fats have been more recently studied, as they were observed to be quite elevated in 4 PA patients after the onset of metabolic crisis. Further observations revealed very high blood levels of these fats in newborn babies who presented in crises, and the level of these fats correlated with their clinical

NUTRITION MANAGEMENT CONCLUSION

condition, more closely than urinary levels of organic acids. But these investigators also found continuously increased levels of these odd chain fats in clinically stable patients. Concern has been expressed in using these methods as measurements of metabolites have not consistently yielded useful information about clinical progress. (Meissner, Clin Chem Lab Med, 2004). Clinics need to find which method will best provide useful information in determining degree of metabolic stability in their PA patients. Besides measuring metabolic stability, there are other important parameters to monitor in PA patients including growth and development, dietary intake, clinical exam, and measurement of other important blood components including complete blood count, measures of protein status such as albumin and prealbumin, and measures of vitamin/mineral status such as vitamin B12, folate, and iron levels.

It is important to avoid constipation as this could aggravate metabolic control. Studies have noted that regulation of bowel movements decreased blood ammonia levels along with lower levels of organic acids in the urine, and increased amounts of carnitine in the blood.

Other Therapeutic Approaches to Help Maintain Metabolic Stability

Constipation can be a fairly common problem in patients with PA, especially when their mobility is limited, if they have low muscle tone, and because a good portion of diet comes from metabolic formulas that do not include dietary fiber. An investigator looked at how many bowel movements 4 PA patients had on a regular diet over a 4 day period. During the following 3 days, while being given the same diets, he gave each of them Senokot syrup, which is a laxative. As expected, he saw a significant increase in the frequency of bowel movements after administering the Senokot. He also noted that blood ammonia levels decreased along with lower levels of organic acids in the urine, and increased amounts of carnitine in the blood. The rate of bowel movement transit time, as measured by a dye added to the patients' diets, had decreased from 33 to 22 hours. The investigator concluded that it was important to avoid constipation as this could aggravate metabolic control. This study was only done over a 7 day period, hence it would be helpful to conduct an investigation over a greater time period to better assess long term impact of treatment. The medication, carnitine, can also work as a laxative, and some clinicians found that by increasing the dose of carnitine can help with bowel management. Since constipation can aggravate metabolic stability, it is important to communicate with the clinic when the individual with PA is having difficulties having regular bowel movements. (Prassad, J Peds, 2004).

Early and Aggressive Treatment Leads to Improved Survival and Outcome

Early detection and more aggressive treatment of babies born with PA is associated with improved survival rates, normalization of growth and nutritional status, and improved developmental outcomes. Metabolic stability has been observed to be easier to maintain as the PA individual gets older. The reason may be because of less frequent illnesses and increased metabolic 'reserves' by the body which eventually can attempt to compensate for the blocked PCC pathway, and use alternate energy sources. Many individuals continue to have problems with their mobility and cognition. Additional treatment options are being studied including organ transplant, growth hormones, and antibiotic treatment, to help improve the outcome of individuals with PA. In the future, there may be the potential for gene therapy, which would completely correct the biochemical defect. But until then, the best approach is to work closely with the metabolic clinic to ensure the most appropriate and adequate diet is provided for management of PA.

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ILLNESS MANAGEMENT FOR PROPIONIC ACIDEMIA

SPECIAL ILLNESS INSERT: KEEP THIS ISSUE AT HAND FOR REFERENCE.

Sandy van Calcar, MS, RD, Metabolic Dietitian Biochemical Genetics Program, University of Wisconsin-Madison

As all parents of a child with PA are aware, knowing what to do during illness is a vital part of management for this disorder. Illness can be very scary for both a child and parents. The most important thing to remember when your child is ill is to contact your metabolic clinic and let them know about the illness. This is especially critical when your child has signs of advanced illness, such as vomiting that doesn't stop, extra sleepiness and/or difficulty walking. If any of these symptoms are present – Call Your Clinic Immediately, or if it is after hours or on a weekend, follow your clinic's policy for emergency management.

What you need to know before your child becomes ill

When your child is first diagnosed with PA, your clinic should provide you with clear information about what happens during illness and what procedures you will need to follow during an illness. Some things to discuss with the staff include:

1. Obtain an "Emergency Protocol". This protocol includes clear directions that can be given to any medical personnel during an illness. Be sure the protocol includes phone numbers to page the "metabolic physician on call" in an emergency.

2. Contact your local MD so he/she understands your child's diagnosis. Your metabolic clinic can send information about PA and emergency management. Some local physicians do not feel comfortable following a child with PA. It is important to know this ahead of time so that you can locate a primary MD that is willing to work with you as well as your metabolic clinic.

3. Find out where you need to take your child to get blood drawn and sent off for quick analysis. Different clinics have different labs that they monitor during illness. You should know what these tests are and develop a plan for getting these labs drawn and analyzed as soon as possible.

4. Learn the process for a hospitalization at both your community hospital and at the hospital associated with your metabolic clinic. Our clinic recommends that families go to the hospital's Emergency Room for evaluation and admission, if necessary. But, different clinics have different procedures for an admission.

5. Order a "Medical Alert" bracelet or necklace so that if your child is in an accident and you are not there, emergency personnel will know your child's diagnosis.

Diet management during illness

Most clinics prescribe a "sick day diet" for kids with PA. The purpose of the sick-day diet is to increase calorie intake and reduce intake of "whole protein." (Whole protein is protein from food that contains methionine, valine, isoleucine and threonine). Both of these steps can help slow protein breakdown that occurs during an infection or other illness.

The amount of whole protein removed in the sick-day diet often depends on how sick your child is. If your child is "just coming down with something" or has a more minor illness, only half of the protein may be removed from the diet. With major illnesses, all whole protein sources are often removed. Your metabolic dietitian can help determine what dietary changes you need to make during an illness.

Initiating a sick-diet can be difficult. When a child with PA is ill, he/she is often not hungry and may be nauseous. If a child will eat, using low protein products and recipes can help. Try to optimize the calorie content as much as possible, so try adding extra margarine to noodles, extra jam on low protein toast and other tricks to add calories to what your child is willing to eat.

Usually, we want the child to continue taking formula (without the whole protein) during an illness if he/she is tolerates it. Providing small volumes frequently throughout the day is best. We also usually recommend that small volumes be given during the night too. Sometimes, alternating formula with juice or other beverages can make it easier to drink formula.

Adding extra calories from sources of simple sugars may also help. Sometimes these are easier for a child to tolerate than formula. Examples here include popsicles, fruit ices or frosting. Sometimes, it is easier to take beverages when ill. Juice (real fruit juice or drinks like "Hi-C"), sports drinks, "smoothies" (juice with fruit and finely crushed ice) or even "milkshakes" made with nondairy creamers may be tolerated. (Taking "milk"-based beverages may be more difficult during illness, but some kids prefer this option).

Adding Polycose® can increase the caloric content of juice or sports drinks. Polycose is a carbohydrate supplement that supplies a quick source of glucose. Polycose is not sweet and doesn't change the taste of what it's mixed with, which can make it easier to tolerate than sugar.

ILLNESS MANAGEMENT FOR PROPIONIC ACIDEMIA CONTINUED

Here are some recipes that we recommend:

Juice:

8 oz juice

2 Tbsp Polycose powder

1/4 teaspoon salt (to provide some sodium)

Gatorade or similar sports drink: (Make sure it is not a low calorie sports drink like Propel®)

8 oz Gatorade

2 Tbsp Polycose powder

For infants and toddlers, we often recommend Pedialyte® with added Polycose. Pedialyte should not be given without Polycose, unless this is recommended by your clinic. Pedialyte is very low in calories (4 Calories/ounce) and will do little to help a child with PA. We recommend the following recipe, which has 20 Calories/ounce. (This is the same amount of calories found in infant formula).

8 oz Pedialyte

1/3 cup Polycose powder

Home monitoring during illness

Some clinics recommend measuring urine ketones during illness. Measuring urine ketones is a quick test that can be completed at home. To measure ketones, Ketostix® strips are saturated with urine. After waiting 15 seconds, the color of the strip is compared to a chart that comes with Ketostix. The darker the color, the more ketones are present. Ketones are produced during illness and, in general, will be greater when your child is acidotic, which indicates poorer metabolic control. Elevated ketones ("moderate" or "large" ketones) should be reported to your clinic.

Collecting urine from an infant wearing disposable diapers can be difficult since these diapers are so absorbent. Cotton balls can be placed in the diaper to collect enough urine. Your clinic may have other tricks for measuring ketones, too.

When to contact your clinic

It is very important to call your clinic to let them know your child is ill. Your clinic's advice is vital to your child's recovery. Some parents feel that they "don't want to bother" the clinic staff. Don't think that way!! Physicians and dietitians in metabolic clinics are trained to help during an illness and we really need to know if your child is ill. We want to know when you put your child on sick-diet, even if your child is "not very sick" or is "just not feeling right".

"Some parents feel that they "Don't' want to bother" the clinic staff. Don't think that way!! Physicians and dietitians in metabolic clinics are trained to help during an illness and need to know when your child is ill"

Depending on the severity of the illness, your metabolic clinic may recommend that you see your local physician to find out "what's wrong" and to start treatment (i.e. antibiotics) for the illness right away. For older children, antiemetics (medications that reduce nausea and vomiting) may be prescribed.

If a visit to the ER or hospital becomes necessary, your child will likely be given IV's (intravenous fluids) to provide calories to help slow protein breakdown. IV's may also contain different electrolyte (like sodium and potassium) or bicarbonate, which can help correct acidosis. Sometimes, these IV's' are enough to help your child feel better so he/she can start drinking formula and eat again. If your child does not feel better, a nasogastric (NG) tube may be placed through the nose into the stomach. NG tubes can be used to give formula and more calories to help slow protein breakdown. Sometimes, especially if your child continues vomiting, an IV amino acid solution may be added to the IV regimen. These amino acid solutions provide a source of amino acids, but do not contain methionine, valine, isoleucine or threonine. The goals of a hospitalization are to slow protein breakdown and ultimately, promote anabolism (rebuilding of body tissue).

It is very important that your child be able to take formula and some beverages/foods before he/she is discharged. We often recommend that IV's not be completely discontinued until the child is able to drink 50% of his formula volume and eat sufficient calories. If your child goes home before he/she is able to take enough formula and calories, then he/she might end up making a return visit to the hospital.

For some kids that get ill frequently or may live far away from a hospital, parents can learn to insert a NG tube to give formula or other calorie sources. It can be easier to give calories by tube than by mouth. For some kids, a G-tube (Gastrostomy tube) may be prescribed. A G-tube is a tube that is surgically placed directly into the child's stomach. G-tubes are often prescribed for kids who just can't take enough calories, even when they are healthy. Formula and other calorie sources can be given through the g-tube during an illness. However, it is important not to use the NG or G-tube if your child is vomiting. Only you and your clinic can determine if either of these tubes would be beneficial for your child.

PAF FUNDRAISING SPOTLIGHT

Check out some of the amazing fundraisers across the country since out last newsletter...

Worldwide- PAF Virtual Dinner & Dance- raised $3360

Georgia- Sheila Buice hosted a Pampered Chef party in honor of daughters Tiffany & Amber- raised almost $1000

South Dakota- Jana Sweetman hosted a boutique evening with friends in honor of her son Ben- raised $6000

Connecticut- Tina Franchi & family coordinated Bianca's Bike Night- $3300 & counting!

New York- Sandy McKillop made business cards "Cans for a cure for Propionic Acidemia Foundation" and with help of friends raised $82 through collection of aluminum cans.

Ohio- Hog Roast & CH Tourney, Pomeroy, raised $3200

-Reayna Tobias asked her friends to donate money to PAF for her 3rd birthday in honor of her friend Gwen- raised $125

-Renee Stewart hosted a backyard July 4th BBQ and asked for donations to PAF- raised $433

-JP Morgan Chase Ice Cream Social- raised $504

-3rd Annual CH Tourney & Tailgate Party- raised over $19,000

The Mouat home in Ohio and the Smith home in Illinois are currently signed on for "Lights of Love" in December of this year. They will be competing for the most "Griswold-like" décor by adding one light for each $1donation to PAF through their respective campaigns. If you're interested in making your home a "Lights of Love" home, contact Jen Mouat at jenmouat@gmail.com or Brittany Smith at brittanydague@yahoo.com.

The Mouat home in 2006 was lit up with over 5,040 lights.

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 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. We are deeply appreciative to Publishers Printing Company for donating the printing costs of the newsletter.*

PA FAMILY DIRECTORY

PAF is compiling an Family Directory that will be distributed to PAF families. If you would like to have your family information included in the Directory, please complete the following form and mail to PAF, 1963 McCraren, Highland Park, IL 60035. Please indicate on the form which information you give permission to include in the Directory. Thank you.

Name - First, Last	Propionic Acidemia?	Gender	Date of Birth	Permission to Include?
Self:_____	YES / NO	F / M	_____	YES / NO
Spouse/Partner:_____	YES / NO	F / M	_____	YES / NO
Child:_____	YES / NO	F / M	_____	YES / NO
Child:_____	YES / NO	F / M	_____	YES / NO
Child:_____	YES / NO	F / M	_____	YES / NO
Child:_____	YES / NO	F / M	_____	YES / NO
Other:_____	YES / NO	F / M	_____	YES / NO

Family Address:_____ City, State, Zip _____ YES / NO
Home Phone: _____ YES / NO
Cell Phone: _____ belongs to:_____ YES / NO
Work Phone: _____ belongs to:_____ YES / NO
Email Address_____ belongs to:_____ YES / NO
Email Address_____ belongs to: _____ YES / NO
Please list if fluent in language(s) other than English_____ YES / NO

I would like to be listed in PAF's mailing list and Family Directory. Information that will be distributed to other families includes general contact information such as name, address, email, phone number, names and ages of children and whether affected or not. By agreeing to be listed, I am willing to let individuals touched by PA contact me for support.

_____ _____
Printed Name Signature & Date

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* _____

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

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PROPIONIC ACIDEMIA FOUNDATION

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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