



ARPKD/CHF Alliance

Autosomal Recessive Polycystic Kidney Disease and Congenital Hepatic Fibrosis

Page 1

Spring 2004

Message from the President

This is an energizing time! A comprehensive research study addressing *all* clinical manifestations of our disease is underway: "Clinical Investigations into Autosomal Recessive Polycystic Kidney Disease and Congenital Hepatic Fibrosis". The ARPKD/CHF Alliance was instrumental in generating the interest for this study. Enrollment for this and the ARPKD/CHF Alliance Repository continue.*

Because of improved prenatal/neonatal care, now more than ever many afflicted with this condition survive and thrive. Nevertheless a great deal of information provided to caregivers and patients about this disorder is outdated. Accurate information is essential for appropriate diagnosis, treatment, and prognosis. The most substantial information and resources addressing our disorder is the ARPKD/CHF Alliance's website (arpkd.org). The ARPKD/CHF Alliance is committed to developing additional website information and raising awareness (see "Professional Educational Project"- on our website).

We appreciate that there has been 20 years of PKD research, however little has changed the perception or long time outcome for those with ARPKD/CHF. Our patient community would certainly benefit from diagnostic criteria and treatment guidelines, as it stands medical care is dependent on local physician and institution. The only at length, easy accessible information addressing medical care for "individualized care" is located on the ARPKD/CHF Alliance's website: "Clinical Care Considerations". Although noteworthy, this is not comprehensive nor "expert consensus driven".

The "NIH Roadmap" recognizes the challenge of basic science findings reaching and assisting the public (see "NIH Roadmap" page 7). When the consumer voice is included in serious ways, through every step of the research

process: how research is prioritized, commissioned, undertaken, evaluated and disseminated, it can generate profound, satisfying results for all involved.

We are eager for research that services the patient, designed to improve quality of life, patient outcomes, quality of medical management, clinical practice and sustained practitioner behavior change specific for this disorder. As patient advocates - that is how we measure success. We are hopeful that with sufficient enrollment, the clinical NIH study will eventually translate into useful clinical information, and in time appropriate treatments will be designed/implemented.

Can you make a difference? Of course. There are multiple ways to get involved. Read through the newsletter and get acquainted. Think of what interests and matters to you most; perhaps you have a special skill or talent you can share with the Alliance, or consider making a donation with the form located on page 15. Imagine if everyone were involved in some way on some level.

The ARPKD/CHF Alliance's light does not go out till everything that can be done, is done for this disorder. "When children's lives are at stake, patience is not a virtue". We will facilitate collaborations with other communities and build coalitions whenever possible; ultimately, our goals focus on "improving the lives of those affected" that is the ARPKD/CHF Alliance's purpose.

Colleen Zak
czak@arpkd.org

Inside this issue:

New Listserv for Electronic Alliance Newsletters—Please Join Us!	2
Presentation of Data and Filing for Orphan Designation, for ARPKD?	4
ARPKD/CHF Awareness Pins	6
Rare Diseases Clinical Research Network	7
ARPKD/CHF Socials	11
Our Supporters in 2003	13
Calling for Photos	14
What you can do to help ...	15
Living with ARPKD/CHF—from a 50 year old...	17

* If you would like additional information on these studies, please contact us, or visit www.arpkd.org. Many thanks to the wonderful staff at Angel Flight, a nonprofit, public charity when upon referral, assists families to the NIH study. Thanks also to NORD (National Organization of Rare Disorders) for featuring the NIH study in their newsletter!

ARPKD/CHF Alliance

P.O. Box 70, Kirkwood, P.A. 17536, Phone/Fax: 717-529- 5555,
Email: info@arpkd.org, www.arpkd.org

Board Members:

Colleen B. Zak, President
Mark Spier, Vice-President
Beth Hall, Secretary
Brian Kopan, Treasurer
Gloria Dengler-Mondaruli
Mark Sanders, Ph.D.

Bereavement Coordinator:

Dawn Chandler

Professional Advisory Board:

Eric Johnson, Ph.D.
Prevention Genetics
Marshfield, Wisconsin

Bernard S. Kaplan, M.D.
Division Chief of Nephrology,
The Children's Hospital of Philadelphia
Philadelphia, P.A.

Kevin E. C. Meyers, M.D.
Assistant Division Chief of Nephrology,
The Children's Hospital of Philadelphia
Philadelphia, P.A.

David A. Piccoli, M.D.
Division Chief of Pediatric Gastroenterology and Nutrition
The Children's Hospital of Philadelphia
Philadelphia, P.A.

Sharon Terry, M.A.
CEO of the Genetic Alliance, Washington, DC
President of PXE International, Inc.

Vicky Holets Whittemore, Ph.D.,
Director, Center Without Walls & Senior Scientific Advisor,
Tuberous Sclerosis Alliance
Washington, DC

THANK YOU!

A special thank you to **Kevin E. C. Meyers, M.D.**, Assistant Division Chief of Nephrology, The Children's Hospital of Philadelphia, Philadelphia, P.A. for providing the ARPKD/CHF Alliance medical review of newsletters and website material.



IMPORTANT NEW LISTSERV Please join us

“ARPKD/CHF Alliance Reporting” is a new listserv for professionals and families. It will provide 1 to 2 page electronic newsletters (hard copies will NOT be mailed out) and announcements effectively and at minimal cost from the ARPKD/CHF Alliance. This is not an interactive listserv where members can post messages. Only the ARPKD/CHF Alliance will post information.

We will raffle off a new and beautiful 4.5 foot square handmade afghan to a joining member in 2004. Please join us by sending an email to info@arpkd.org, in subject area, type: New listserv. It's that simple.

Thank you to Susan Walushen, creator and donator of afghan.

If you have any questions, please call or email the ARPKD/CHF Alliance at 717-529-5555 or email at info@arpkd.org



Special points of interest in upcoming newsletters:

- Diary and power point presentation of NIH visit
- Additional Testimonials of NIH study
- A Tribute to Fifty Years of Transplantation
- Understanding the National Institutes of Health
- How to find a Genetic Counselor
- Interpreting Lab work
- ARPKD/CHF research
- ARPKD/CHF Alliance Strategic Goals

PASSAGE OF GENETIC NONDISCRIMINATION LEGISLATION

WASHINGTON, DC. Genetic Alliance applauded the United States Senate's passage of legislation today banning genetic discrimination in health insurance and employment.

Health insurers would be prohibited from using predictive genetic information to deny, refuse, renew or change the terms, premiums or conditions of insurance coverage.

Mary Davidson, Executive Director of the Genetic Alliance, hailed the Senate's passage of the Genetic Nondiscrimination Legislation "a fitting first civil rights legislation for the 21st century." Davidson commended the Senate's leadership for building consensus on both sides of the aisle to pass legislation that will protect all Americans from genetic discrimination.

S. 1053, the Genetic Information Nondiscrimination Act, would prevent health insurers and employers from denying an individual health coverage or employment on the basis of his or her genetic predisposition to a disease. Health insurers would be prohibited from using predictive genetic information to deny, refuse, renew or change the terms, premiums or conditions of insurance coverage. Employers would be barred from using this genetic information in the decision-making processes of hiring, firing, promotions or other job-related issues.

With Senate passage of the legislation, the focus now shifts to the US House of Representatives. In 2001, two House committees held hearings on genetic discrimination; however there has been no House activity over the past two years.

Sharon Terry*, President of the Genetic Alliance, called on the House to take action immediately on this important legislation: "Genetics is about all of us and knows no political boundaries. We must protect Americans from the misuse

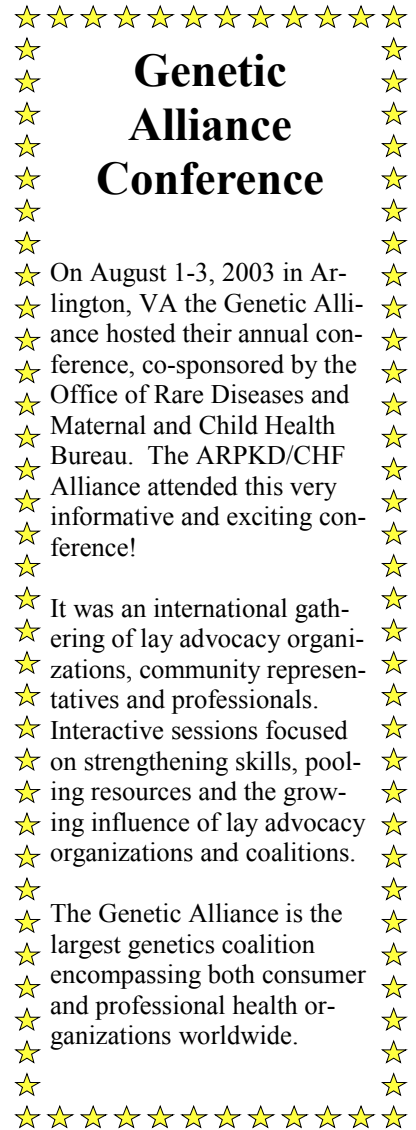
of genetic information. This will give people the security to seek needed healthcare and ensure that genetic advances are used to everyone's benefit."

In the House, over 300 organizations* including the Genetic Alliance and many of its coalition members, have endorsed H.R. 1910, Genetic Information Nondiscrimination in Health Insurance and Employment Act, which is sponsored by Rep. Louise Slaughter (D-NY) and has strong bipartisan backing. Nearly 50 Republicans have signed on as well as key GOP leaders.

Diane Baker, Vice President for Professionals of the Genetic Alliance, asserted, "Genetic advancements will open new doors to medical treatments and improved public health. For these benefits to be realized, people need to feel secure about the consequences of seeking health care and participating in research studies without fear about how their genetic information will be used. This legislation is key in providing reassurance to the public that their genetic information will not be misused by their health insurers or employers."

**Sharon Terry is an ARPKD/CHF Alliance Professional Advisory Board Member.*

**The ARPKD/CHF Alliance endorsed this bill.*



Genetic Alliance Conference

 On August 1-3, 2003 in Arlington, VA the Genetic Alliance hosted their annual conference, co-sponsored by the Office of Rare Diseases and Maternal and Child Health Bureau. The ARPKD/CHF Alliance attended this very informative and exciting conference!

 It was an international gathering of lay advocacy organizations, community representatives and professionals. Interactive sessions focused on strengthening skills, pooling resources and the growing influence of lay advocacy organizations and coalitions.

 The Genetic Alliance is the largest genetics coalition encompassing both consumer and professional health organizations worldwide.



**Have you moved or are you going to move?
Please notify of us of address changes.
Returned newsletters are costly and benefit
no one.**

**If you would like to like to receive newsletters via email,
please send your email address to info@arpkd.org**

**We encourage you to share this newsletter with other
ARPKD/CHF families and the professionals who care for
them.**

AVI BioPharma Announces Presentation of Data and Filing for Orphan Designation

11/13/2003--AVI BioPharma, Inc., announced the presentation of data from a preclinical study describing the use of a novel NEUGENE(R) antisense compound in a mouse model of the childhood form of polycystic kidney disease (PKD), also referred to as autosomal recessive PKD (ARPKD). In addition, AVI announced that it has filed an application with the U.S. Food and Drug Administration (FDA) to obtain orphan designation for another NEUGENE compound, AVI-4126, for the potential treatment of patients with ARPKD.

In the study, a novel AVI compound inhibited PKD1* gene expression and resulted in the reduced size of renal cysts and some preservation of kidney function. Mutations in the PKD1 gene are considered to be the major cause of PKD.

"AVI has experience with both forms of PKD, the most common genetic disease. The data presented this week in ARPKD add to our overall understanding of PKD," said Dr. Patrick L. Iversen, senior vice president of research and development at AVI. "The data we have accumulated from our previous studies evaluating NEUGENES in the childhood form of PKD established the basis for the orphan designation filing."

About AVI BioPharma:

AVI BioPharma develops therapeutic products for the treatment of life-threatening diseases using two technology platforms: third-generation NEUGENE antisense drugs and cancer immunotherapy. AVI's lead NEUGENE antisense compound is designed to target cell proliferation disorders, including cardiovascular restenosis, cancer, and polycystic kidney disease. In addition to targeting specific genes in the body, AVI's antiviral program uses NEUGENE antisense compounds to target single-stranded RNA viruses, including West Nile virus, SARS, coronavirus, calicivirus and hepatitis C. AVI's second technology, AVICINE(R), is a therapeutic cancer vaccine with late-stage trials planned for the treatment of pancreatic cancer. More information about AVI is available on the company's Web site at <http://www.avibio.com/>.

***Editor's Note: PKD1 relates to Dominate PKD or ADPKD**

What is the Orphan Drug Act?

There are many diseases and conditions in the United States that are considered rare in the United States, defined as those afflicting fewer than 200,000 people in the United States. Drugs for these diseases and conditions are commonly referred to as "orphan drugs", because so few individuals are affected by any one rare disease or condition.

The Orphan Drug Act was passed twenty years ago to promote research into rare disorders. It uses several enticements to spur drug makers to research treatments for orphan diseases. Incentives include tax credits, grants and a seven-year guarantee of market exclusivity for resulting drugs. The Act also provides for a clinical research grants program whereby researchers could compete for funding to conduct clinical trials that might support the approval of these drugs for rare diseases.

What is the difference between an orphan grant and an orphan designation?

The orphan designation process is the mechanism by which sponsors of drugs and biologics for rare diseases qualify for the tax credit and marketing exclusivity incentives of the Orphan Drug Act. The Office of Orphan Products Development also administers a clinical research grants program, whereby researchers may compete for funding to conduct clinical trials to support the approval of drugs for rare diseases.

Can an orphan drug application be submitted anytime in the development process?

Under the law, application for designation as an orphan product can be made any time prior to the filing of an application for approval to market the product.

Information obtained from "OOPD Frequently Asked Questions" <http://www.fda.gov/orphan/faq/>

New Guidelines for Blood Pressure Readings

Normal isn't normal anymore. Blood pressure "normal readings" have been redefined. What used to be considered normal last year, is abnormal this year. Updated evidence-based guidelines, released by the National Heart, Lung and Blood Institute (NHLBI) were published May 21, 2003 in the *Journal of American Medical Association*. These new recommendations for managing abnormal BP, is in the NHLBI's *Seventh Report of the Joint National Committee on Prevention, Detection, Evaluation, and Treatment of High Blood Pressure* (the JNC 7, for short—for more information go to: www.nhlbi.nih.gov/guidelines/hypertension).

Some tips on how to take a proper BP reading:

- Patient should be seated, with feet down.
- Use a calibrated machine with a cuff that encircles at least 80% of the arm; arm should be at heart level.
- Take the average of two blood pressure readings
- Two or more visits, a total of four readings are usually needed before a diagnosis of abnormal BP can be made.

New Growth Charts

In May 2000, the Centers for Disease Control (CDC) released new growth charts, which are more representative of the current U. S. population.

Who created the new growth charts?

The Centers for Disease Control, the Department of Health and Human Services, and experts from academic institutions across the country.

What is different about the new charts?

The 2000 growth charts appear similar to the old growth charts, however, two new charts will be added including Body-Mass Index (BMI), for boys and girls aged 2 to 20. The new charts are more representative of all racial and ethnic groups, and represent the combined growth patterns of breastfed and formula-fed infants.

The new growth charts will continue to assist health professionals in identifying those children at risk for underweight or overweight. For more information please visit : <http://www.cdc.gov/nchs/about/major/nhanes/growthcharts/clinical>

Acidosis - by Gloria Dengler-Mondaruli

My son, Michael, age 5, was diagnosed with ARPKD several days after birth. His condition has been stable and he is currently taking Enalapril and Furosemide to control his blood pressure. Since he was 6 months old, the carbon dioxide level in his blood was low and he was considered to have metabolic acidosis. [According to one report...metabolism of the foods we eat produces acid, which normally leaves the body in the urine. When the kidneys are not functioning properly, this acid accumulates in the blood. It does not cause any symptoms, but it can slow growth, and cause calcium to leave the bones, weakening them. Medication is successful in "neutralizing" this acid build-up; periodic blood tests are necessary to measure the acid level in the blood and adjust the medication.] Succinctly, the doctor said if the acid level goes too low, it will start to eat at the bones, leaving him with growth challenges.

When he was a baby, I remember trying to give him a mixture of baking soda and water in a syringe, as the doctor prescribed. He got sick many times over and I refused to continue a remedy with that kind of reaction. That was soon replaced with Bicitra three times a day which I mixed in his bottle. His levels remained low, although I will admit there were times I was remiss in giving it to him. At age 4, I stopped giving him the bottle and lost the ability to hide the bicitra in his bottle. Adding the medicine to his sippy cups didn't work nor did trying to shoot it into his mouth with a syringe. The doctor suggested giving him crushed sodium bicarbonate tablets which we mixed with all kinds of juice to hide the awful taste. No luck. The struggles continued. We even went as far as consulting with a compounding pharmacist who gave us a container of granulated sodium bicarbonate, a bottle of glycerin emollient and a bottle of cherry syrup. Our kitchen soon became a laboratory and our son became the experiment. God bless my husband for taking over the role of mad scientist. After many attempts at trying to get the mixture into his body, we gave up.

Just by accident, little Michael took a liking to drinking Gatorade on a regular basis. Without thinking, I gave it to him whenever he asked for it just to get him off his milk habit!! His carbon dioxide jumped 3 points from 18 to 21 (normal range is 21-33) in a short time without supplementation (meaning bicitra or sodium bicarbonate). One night while at the store to pick up more "Gatorade", I happened upon "PowerAde", remembering a note forwarded to me from a woman whose daughter's carbon dioxide was low. She recommended drinking at least 32 ounces of PowerAde. Well, I compared the ingredients and, low and behold, they almost matched. Go figure! I'll continue giving my son Gatorade with the hope that his next blood workup will see more good results!

Please be informed that I am not endorsing any of the aforementioned medications, treatments, products or opinions in this article. This information is intended only to keep you informed. You should always discuss any diagnoses, treatment or medications with your physician.

Taylor Grace—9 months old by her mother Sherri

This disease is so confusing. I did not find out until 10/30 of last year when I was six months pregnant. I spent almost a month in the hospital for low amniotic fluid, doctors claimed she was urinating on the ultrasound, but it just kept disappearing. They spotted a cyst on her kidney and sent me to a high risk OB which never once mentioned to me about any kind of kidney disease. He told me that the cyst did not seem to be a factor but her hole in the heart was. So we also saw a cardiologist prior to delivery. I spent a month in and out of the hospital and they gave me steroid shots to boost lung development.

They took her five weeks early and she was 5 lb, 14oz; her heart was the primary concern. She spent ten days in NICU where they called in a Genetics Specialist. They told us about the possibility of this disease but felt that since she was doing OK that she had 'adult' PKD and said to follow-up with doctors later. In May we had her open heart surgery, in which she developed a UTI prior to surgery and again after surgery.

Her GI & pediatrician did not want the surgery done until we verified that her kidneys were functioning properly due to the dangers of the surgery. She has

NEVER not once acted like a sick baby. Her BP is normal every time we have been to the nephrologists. Her labs are great so they took her off the Bictra and we cut in half her Lasix to .3ml. Possibly next lab we will do away with them. This would mean no medications at all.

They still say she has characteristics of either ADPKD or ARPKD but pretty certain it is ARPKD/CHF even though there is no history anywhere in our large families. We have not had an ultrasound since July so in a week or two we will see if the cysts have gotten any larger. Her nephrologist told me we could start doing labs every 3 months and ultrasound every 6 months since things with her are normal. I know that they will not always be normal but I have to hope it is a very, very long time before any symptoms develop and the research has

found a cure by then.

This past Monday Taylor was nine month old and weighs 21lbs & 7oz. and trying to move that chunky monkey body of hers around. She is right on with normal development. I look back now almost being a year since my life turned so upside down and we had so many obstacles to overcome yet she is defying the odds every day and showing us what a true gift she is! I just wanted to pass on some good news and to tell you all to NEVER to give up hope, miracles do happen. My Taylor is testimony to that!!! I pray for all you every single day, God bless.....

Take care,

Sherri, FL

(Brandon 11 and Chase 4—both clear, Taylor Grace 9 months old with ARPKD/CHF)

Awareness Pins for ARPKD and CHF - by Denise Tarabanovic

As I read e-mails from parents and family about doctors not giving any hope when an ARPKD diagnosis has been given it makes me really upset. I realize how lucky I was that the doctors who took care of Clayton after birth knew about this disease and gave us hope. So how do we get the doctors, hospitals and general public aware of this disease? Well it's not by looking in medical books or most things on the internet, as most sites portray children dying right after birth.

Though we do have our ARPKD angels in heaven looking out for us, this is not always the norm anymore. Children are

surviving more and more. So how do we bring awareness for our condition? Well



I thought an awareness ribbon would be a good way to get people asking about what the color means. And in doing so explaining what the disease is and of course direct-

ing them to a wonderful web site: www.arpkd.org. So I spent hours on the internet looking up colors that weren't used by another cause. What a task that

was! I found a few colors and it was agreed upon that **mint** would be the awareness color for ARPKD/CHF. **I encourage everyone to wear a mint colored awareness ribbon.** I can't tell you how many people have asked about my ribbon.

I bought the awareness pins from 'The Ribbon Factory'. Their website is www.ribbonfactory.com. They only sell the ribbons with safety pins that you must attach yourself. The price for one bag—250 count, is \$20. I'm still looking for a company that will make other items such as pins, stickers etc. I'll let you know what I find.

NIH Roadmap for Medical Research

In a move to transform the nation's medical research capabilities and speed the movement of research discoveries from the bench to the bedside, National Institutes of Health (NIH) Director Elias A. Zerhouni, M.D., laid out a series of far-reaching initiatives known collectively as the NIH Roadmap for Medical Research.

To improve human health, scientific discoveries must be translated into practical applications. Ideally, basic research discoveries are quickly transformed into drugs, treatments or methods for prevention.

To accelerate and strengthen the clinical research process, NIH Roadmap initiatives will re-engineer the clinical research enterprise by adopting a systematic infrastructure that will better serve the evolving field of scientific discovery. This effort, which complements the other initiatives that comprise the NIH Roadmap, will provide the necessary foundation for advancing basic and clinical research. With the NIH Roadmap in action, investigators will be better poised to translate basic discoveries into the reality of better health for our nation.

To improve human health, scientific discoveries must be translated into practical applications. Ideally, basic research discoveries are quickly transformed into drugs, treatments or methods for prevention.

Major NIH Roadmap Themes: New Pathways to Discovery, Research Teams of the Future, Re-engineering the Clinical Research Enterprise. For much more information see: <http://nihroadmap.nih.gov>

NIH ESTABLISHES RARE DISEASES CLINICAL RESEARCH NETWORK

"The network will facilitate increased collaboration and data sharing between investigators and patient support groups working to improve the lives of those affected by these diseases and potentially prevent or eliminate these diseases in the future," said Stephen Groft, Pharm.D., director of NIH's Office of Rare Diseases.

The National Institutes of Health (NIH) announced today the establishment of the Rare Diseases Clinical Research Network. With \$51 million in grant funding over five years from several NIH components, the network will consist of seven Rare Diseases Clinical Research Centers (RDCRCs) and a Data and Technology Coordinating Center (DTCC).

"Funding research on rare diseases is a vital aspect of the NIH mission," said NIH Director, Elias A. Zerhouni, M.D. "By encouraging cooperative partnerships among the investigators at these centers, we hope to accelerate the development of diagnostics and treatments that will benefit these important patients."

"Because of the small number of affected patients in any one location, rare disease research requires the collaboration of scientists from multiple disciplines and the capacity to share access to geographically distributed national research resources and patient populations," said Judith L. Vaitukaitis, M.D., director of the National Center for Research Resources (NCRR), one of the NIH funding components. "With a collaborative approach, the network will focus on identifying biomarkers for disease risk, disease severity and activity, and clinical outcome, while encouraging development of new approaches to the diagnosis, prevention, and treatment of rare diseases."

"The network will facilitate increased collaboration and data sharing between investigators and patient support groups working to improve the lives of those affected by these diseases and potentially prevent or eliminate these diseases in the future," said Stephen Groft, Pharm.D., director of NIH's Office of Rare Diseases. "In addition, knowledge about rare diseases may offer leads for scientific advancement in other rare diseases and in more common diseases."

This network supports the re-engineering of the clinical research enterprise component presented recently in the "Roadmap for Medical Research" by Dr. Zerhouni. Each research center consists of a consortium of clinical investigators **partnering with patient support groups** and institutions within and outside of the United States that have agreed to work together studying a group of rare diseases. In addition to fostering collaborative research, the RDCRCs will train new investigators for the represented rare diseases and provide content for a public Web site on rare diseases research.

Approximately 25 million people in the United States are affected by an estimated 6,000 rare diseases or conditions. The term "rare (or orphan) disease," as defined in the Orphan Drug Act, is a condition affecting fewer than 200,000 in the United States or a disease with a greater prevalence but for which no reasonable expectation exists that the costs of developing or distributing a drug can be recovered from the sale of the drug in the United States.

Karen's story—from a 23 year old living with ARPKD/CHF

I guess I will begin when I was 5 and my pediatrician noticed an enlarged spleen. He never really paid much attention to it, except to tell my parents to be cautious in case I was ever in an accident. Besides that, I was a healthy child. I had a few colds here and there, experienced the flu, but no major problems. My senior year in high school was when everything changed. I was a student athlete, participated in 3 sports. My best sport was swimming. I even had a few colleges interested in me. About 2 months into the season I started to have sharp pains in the spleen area. The coaches sent me to the athletic trainers who thought I had a broken rib. I informed them of my enlarged spleen and they said I might want to see a doctor.

Within a week I was in and out of 3 or 4 different doctor's offices until finally they admitted me into the hospital. I was in the hospital for about 5 days. My spleen was so enlarged that 10 different doctors came just to feel my spleen. I felt like a circus attraction! They ran CT scans, MRIs, ultrasounds and blood work trying to figure it out; the only thing they could see was abnormalities with my liver. Their last resort was a liver biopsy. Their first 2 attempts with a needle failed because they could not get anything but scar tissue. Finally they used a carving knife and took out a nice chunk! When everything was said and done, the final word was CHF. In 1997 they could not explain much about the disease. All they told me was that it was very rare, and it came from recessive genes. I had a brief encounter with a hepatologist who told me that I would have to be monitored and I would be a high risk pregnancy.

I tried researching the internet but I found nothing. I went to liver disease support groups in search of information and they had never heard of my disease. Finally I began to feel better and I gave up on trying to find an explanation. I went back to school, and swimming (unfortunately I never had much of a comeback) and finally graduated high school. I went to an out of state college my first year. I got sick, and was hospitalized for 3 days. After that, I transferred back to my home state Indiana. It seems that once a year I get sick. They have quit hospitalizing me for more than a day because there really isn't anything they can do except alleviate the discomfort with pain medication. The only regular medication I take is vitamin K (for clotting).

(Continued on page 10)

The ARPKD/CHF Alliance would like to thank the Bereavement Task Force, for their input and contributions to the Alliance website:

Dawn Chandler-Chair, Suzanne DiMarzo, Tracey Emanuel, Kelly Fujii, and Kristen Neary

"The pleasure in living is in helping" ~ Professor Henry Barnes, in "Apartment for Peggy"

Pregnancy and Infant Loss Remembrance Day is October 15th each year.

This is a day of remembrance for all babies who have died too soon, from early pregnancy loss thru infant death. Every life, even the tiniest and shortest deserves to be acknowledge and remembered. The parents of these children never forget.

For more information: <http://www.pain-heartache-hope.com>

Chloe Fujii was born September 14, 2002 and lived for 13 1/2 hours. Her family feels blessed to have had that time with her. In memory of her daughter, Kelly S. Fujii held a raffle fundraiser with family and friends that raised \$725.00. Thank you Kelly for remembering the ARPKD/CHF Alliance.

The Story of Ezra Quail



Hi, my name is Denelle Quail. I live in Eden Prairie, Minnesota with my husband Shane and my 2 year old son Ezra. Ezra was born with ARPKD. When I was pregnant I was diagnosed with gestational diabetes and my doctor asked for an ultrasound to be done when I was 35 weeks along. My husband and I went to the ultrasound and thought everything was fine so he left to go bike riding while I met with the doctor afterwards.

My doctor immediately asked where Shane was and I told him that he had already left. He said that he had terrible news and he wished my husband were there with me. I started to panic. He told me that Ezra's kidneys were 3 times the size they should be and that I had no amniotic fluid left. He was going to set us up with a difficult pregnancy specialist within the next few days to see what was going on.

We waited for 3 days before seeing her. She was a very cold doctor. She did the ultrasound and told us that our baby was going to die shortly after his birth. She said it looked like he had ARPKD and that she doesn't know of any success stories with this disease. She told us to plan for his funeral and to expect him to die immediately after he was born. She said I had to give birth to him within the next few days otherwise he was going to die inside of me. We were devastated. We went home and cried and cried. We had no idea why or what was going on.

I called my doctor begging for a C-section so I could be knocked out during the birth. I just wanted it all to be over with. My doctor refused to do a C-section knowing that I would fall into a deep depression if I didn't get to see my baby. We waited 5 days be-

fore I was induced. I took all of the clothes for Ezra and packed them. We were planning on returning his stroller and car seat. It was terrible.

I woke up the day I was going into the hospital to have Ezra and I turned on the television. At that moment I watched the second plane crash into the World Trade Center. What a day to give birth to my baby who was going to die. I thought the world was ending. We had to go to the hospital on September 11th, 2001 and start the induction. It was such a terrible day. The doctors kept giving me sleeping pills to help me sleep and forget about what was going on. I started to contract at 4 a.m. on September 12th. I had a fairly easy labor with the help of a lot of painkillers. My doctor didn't want me 100% coherent for the delivery.

With 3 pushes Ezra Daniel Quail came into this world at 3:32 p.m. crying and peeing all over the doctor. I thought it was Ezra's way of saying 'ha ha' my kidneys ARE working! Well he was breathing fine. The doctors told us not to help him breath in any way, that we should just let him die. So being the young, vulnerable parents that we were, we agreed.

The doctors told us not to help him breath in any way, that we should just let him die....They told me NOT TO FEED HIM!!!! They said it would hurt his kidneys more and it would prolong his pain and suffering.

Thank God Ezra's lungs were alright. He was breathing, crying and peeing everywhere. He was 5 pounds 8 ounces and 18 inches long. He was baptized 10 minutes after he was born. We had all of our family

and some friends in the room 5 minutes after he was born. Well throughout the hospital stay Ezra was still with us. The doctors continued to tell us that he was going to die. They told us that he would die sometime in the next few days. They told us to take him home and wait. I asked for a bottle in the hospital to feed him since my milk wasn't in and he was acting hungry. They told me NOT TO FEED HIM!!!! They said it would hurt his kidneys more and it would prolong his pain and suffering. I refused to listen to

them and demanded them to bring me something. I thought, they don't starve cancer patients, even though they are going to die. I was going to make my beautiful baby as comfortable as I could until his last breath. They brought me a syringe and sugar water to feed him. He was too tiny to suck and couldn't keep formula down.

We took him home 2 days later and waited. We continued to feed him with a syringe. He learned to suck on a special nipple a few days later. I didn't breast-feed because the nurses didn't show me how. Ezra wasn't circumcised because they said he was going to die. They didn't even show me how to clean his umbilical cord. Well 2 weeks of holding him every second of the day, a doctor called me.

I was literally at that moment sitting on my couch holding Ezra, watching him sleep and thinking, why was he going to die? He looked so perfect and he acted like a normal baby. Well the phone rang. It was a heart specialist that didn't give up on Ezra. My Gynecologist had contacted him to get a hold of pediatric nephrologist and tell them about Ezra. The words that came out of his mouth were "Ezra is going to be in your lives for a very long time." I didn't believe him. I had lost all trust in doctors. He kept repeating it to me and told me a kidney specialist would be calling me that day. I called everyone I knew.

(Continued on page 10)

(Continued from page 9, Ezra's story)

My husband didn't believe the doctor. My parents were ecstatic. My in-laws were amazed! Everyone was praying for Ezra.

That night a doctor called us and explained everything about ARPKD. We made an appointment and saw them a few days later. They did blood work on Ezra and afterwards came into the room saying they had fabulous news, Ezra's kidneys were working 100%!! We took him home and he was back at the doctors 2 months later his blood pressure was through the roof. They immediately put him in the hospital. He was there for 6 days. They sent him home on 3 medications and a blood pressure machine.

He was good for a while and then he was sick with bronchitis and ear infections for about 8 months. He was back in the hospital when he was 7 months old for blood pressure again. His heart was a little enlarged due to the blood pressure. He was there for 6 days again and finally on the right meds. We moved and took him out of daycare. I take care of our nephew 3 days a week so Ezra gets to be with me all of the time.

He has only been sick twice in a year. Compared to being sick his first entire year and not growing. Now he was starting to grow and crawl and walk!!! He is 2 now and doing great!!! His kidneys are still doing really good and his blood pressure is awesome. They just increased his BP medication and it is working like a charm. He is the light of our lives. He is our little miracle that keeps on fighting. The name Ezra means to help people, I hope his story will help people dealing with this same disease.

My advice to all parents dealing with this, speak up, be your own voice. Do your research and always ask



questions. If they don't have answer find out for yourself. You are the one that knows your child best keep the doctors educated.

Ezra is such a joy. He takes medicine twice a day and goes to get his BP and blood work checked twice a month. He sees the kidney doctor every 6 months. There is hope, there are happy endings with ARPKD. I hope this story has helped someone today, I know many people have helped me with their stories. I thank you all who put this website together.

Thank you for hearing our miracle story.

~ Denelle, Shane & Ezra Quail.

If anyone would like to contact me please do so at: denellebla@msn.com

"Any annoyances, frustrations, or challenges the doctors endure are magnified a hundredfold by those the patients experience."

Nicholas L. Tilney, MD,
"Transplant: From Myth to Reality"

If you would like to share your story/experience, please send it to: editor@arpkd.org or info@arpkd.org

(Continued from page 8, Karen's story—a 23 year old with ARPKD/CHF)

Surgeries were discussed but dismissed thinking it would cause more harm than good. As far as other problems - I get sick with the common cold more than most and I keep it for weeks rather than days. My spleen is usually always tender, it is usually worse during menstruation. When my platelets fall I get nosebleeds and my clotting is always a little slow. Sometimes I go through stages of extreme fatigue. I have to watch my stress level because that is when I get really sick. I guess that brings us to today.

I had to switch to the University's insurance plan and change doctors. I informed them of my medical history and they wanted to do tests other than blood work because it had been a few years. I had a CT scan in October and it showed large varices in my esophagus and a cyst in my ovaries. Upon further testing they decided the varices had progressed but not to the point of needing the band technique. This new doctor still wants another biopsy performed. The cyst they found was actually a collection of about 12oz of fluid (most people carry about 2-3 extra oz) due to my portal hypertension. My OB/GYN told me we need to watch this so it does not turn into ascites. This is all I know currently. Unfortunately I no longer have medical coverage. My new provider will not cover "preexisting conditions" until after one full year. We already have racked up quite a bill already.

These last few months have been hard dealing with doctors, insurance agencies, and being a full time student. I found the ARPKD/CHF Alliance website during one of my moments of frustration and thank God I did. I will be going to DC in March for the NIH research study and hopefully they can answer all these questions I have been carrying with me for the past 6 years.

~ Karen W.