ARPKD/CHF Alliance

Autosomal Recessive Polycystic Kidney Disease/Congenital Hepatic Fibrosis

Welcome, Families & Friends

By Colleen Zak

Dear ARPKD Families and Friends,

Have you ever thought back on the choices you've made in life? When you think about it, we are the sum total of all the choices we ever made. Our destiny is not a matter of chance, it is a matter of choice, and some of our personal choices have a profound effect.

Last year a group of ARPKD parents came together with a vision: to improve the lives of those affected by ARPKD/CHF. They agreed to specific goals to the dual organ disease [Autosomal Recessive Polycystic Kidney Disease (ARPKD) and Congenital Hepatic Fibrosis (CHF)] were needed, and they made an exciting and life changing choice. An organization specific to ARPKD and CHF was established: the ARPKD/CHF Alliance was founded and has public charity 501(c)(3) nonprofit status. Board Members and Officers are parents to children affected with ARPKD and CHF, so we are naturally committed to our mission, purpose and patient outcomes.

The ARPKD/CHF Alliance has a unique and special purpose. We are neither a duplication nor an overlap of other organizations and are unparalleled in mission: to educate, advocate, support and advance research specific to Autosomal Recessive Polycystic Kidney Disease (APRKD) and Congenital Hepatic Fibrosis (CHF). Our view and measurement of success is based not just on treatment/cure, but also on improving the lives of those affected by ARPKD and CHF. For those living with the disorder, normalizing and easing life experiences is paramount, while those who have endured the death of an infant or child, at the very least, need bereavement resources and information.

ARPKD is unlike any other cystic disease. There is a distinction between ARPKD and other cystic kidney diseases: CHF is ALWAYS present to some degree. No other organization has focused on or provided a catalyst for understanding CHF. There is a wealth of ARPKD and CHF questions to which we have no answers or understanding; little is known about the disease course and we have no parameters or measurement to determine clinical sequence, symptom presentation, severity range or patient outcomes. Inaccurate, misleading information continues to be disseminated to families, particularly to expectant and newly diagnosed parents.

Identification of the ARPKD/CHF gene early this year by Dr. Peter Harris and his colleagues at the Mayo Foundation in Rochester, Minnesota, is one of the *first* basic steps in understanding the molecular, cellular, biochemical and developmental mechanisms by which ARPKD & CHF occur. This gene is predicted to encode a large protein – fibrocystin, which is not the same as polycystin 1 and 2, associated with the ADPKD genes.

There are no medical based guidelines or references for clinical care of patients with ARPKD/CHF. One of the Alliances goals is to develop a reference point in which medical professionals, patients and parents can access such information to help facilitate appropriate care options/considerations or

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Our Mission:

To educate, advocate, support and advance research specific to Autosomal Recessive Polycystic Kidney Disease (ARPKD) and Congenital Hepatic Fibrosis (CHF)

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ARPKD/CHF patients. We have established the ARPKD/CHF Alliance's Blood Bank Program, a centralized resource open to research projects demonstrating merit, not exclusively controlled by a single researcher or group of researchers, but owned, managed, and funded by the ARPKD/CHF Alliance, persons it directly affects, thus making possible maximal scientific cooperation and dissemination of scientific information. This has generated an impressive invitation to present our registry at the "PKD Strategic Planning Meeting", sponsored by the National Institute of Diabetes and Digestive and Kidney Diseases; please consider your own participation in this registry (see "ARPKD/CHF Alliance Sponsored Research", page 3 for more information).

There are dramatic visual records demonstrating that research can be accelerated, resulting from patient/parent involvement, even for rare disease (size never kept a mosquito from being effective!). Our time factor is critical, we do not blindly accept the conventional view that science can't be focused or pushed along. It is imperative, as concerned parents and patients, to question and stimulate progress; we share ownership and responsibility for success.

Research strategy, data collection, analysis and findings will improve quality of life, improve detection and treatment of ARPKD/CHF, assist in educating healthcare professionals and allow parents to become the best advocates their children have.
It won't be easy, will take hard work, time and energy, requiring commitment, but we won't settle for less.

Sincerely,

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ARPKD/CHF Alliance Sponsored Research

The ARPKD/CHF Alliance is offering the opportunity to participate in a research project to provide a better understanding of Autosomal Recessive Polycystic Kidney Disease (ARPKD) and Congenital Hepatic Fibrosis (CHF).

The purpose of this study is **twofold:**
1) To assist in identifying the molecular biology factor(s) causing ARPKD and CHF.

2) To more accurately describe the disease process, course, progression and outcomes, and its impact on affected individuals.

We are collecting blood samples from individuals affected by this disease, their immediate blood relatives, and in particular affected siblings. This blood may help us to understand the genetic cause and factors causing Autosomal Recessive Polycystic Kidney Disease and Congenital Hepatic Fibrosis.
The ARPKD/CHF Alliance's Blood Bank Donor Program provides a *centralized storage and research distribution to facilitate worldwide investigation* into the genetic causes of the Orphan Diseases Autosomal Recessive Polycystic Kidney Disease (ARPKD) and Congenital Hepatic Fibrosis (CHF). This Bank is unique; it is the only international collection and public source of genetic material available to ARPKD & CHF researchers/research projects approved by the ARPKD/CHF Alliance Review Subcommittee. Researchers have limited time and resources; we provide coordination of retrieval, storage, and distribution of samples. The ARPKD/CHF Alliance's Blood Bank will not be restricted or exclusively controlled by a single scientist or group of researchers, but owned, managed, and funded by the ARPKD/CHF Alliance, by the people it directly affects, available to any number of research projects demonstrating merit; making possible the opportunity of maximal scientific cooperation, collaboration, and dissemination of scientific information as rapidly as possible. This enables and promotes a time sparing approach to research, and allows us to focus on what is needed to improve the quality of life for the ARPKD/CHF population, with expediency and efficiency.

We offer the opportunity to complete a self-administered survey, to provide a better understanding of the disease process, course, progression and outcomes, its impact on affected individuals; in order to better evaluate the relationship between ARPKD and CHF, family and genes. This will provide many important details. Our basic understanding of ARPKD and CHF affecting the population is limited, little is known about the clinical course. Previous estimates of true population occurrence, and morbidity are limited by sample size, do not include quality of life issues that have paramount importance to families, and lack the complete range of experienced clinical symptoms and complications. There is a gamut of symptoms, with a wide range of severity, without any understanding of why some individuals die shortly after birth and others live into adulthood. There is a complete absence of parameters or measurements available to professionals and nonprofessionals in determining patterns, sequence, or patient outcomes.
Further study and focused attention is needed to better understand the disease process at the molecular, cellular, and human level, from the prenatal phase into adulthood, this will have paramount importance in better understanding the condition and potential treatment.

For additional information and details, please contact the ARPKD/CHF Alliance at 717-529-5555 or info@arpkd.org

Train the Trainer Workshop

March 8 to 10, 2002

Recognizing the importance of empowering consumer voices to participate in public policy decision-making, the Genetic Alliance and the Genetic Services Branch of Maternal and Child Health Bureau co-sponsored a Train-the-Trainer workshop on March 8 to 10. Colleen Zak from the ARPKD/CHF Alliance was in attendance. This three-day interactive and dynamic program built the skills and knowledge that participants need to enable their own groups to become effectively involved in local, state and national policy advocacy. Consumer organizations gathered at the Kellogg Conference Center on Galludet University's campus in Washington DC. Program events encouraged networking and coalition building among consumer leaders and across the specific interests of individual disease groups. Training materials are posted on the Genetic Alliance website so that other consumer organizations can benefit from and join efforts to build an even stronger consumer coalition; more information can be found at: www.geneticalliance.org.

The Roberts Family Story

By Julia Roberts

Meet Quinn and Gage (pictured below). They are typical children. Quinn is starting to show her personality and Gage is testing his limits. They both have ARPKD.

On September 29, 2001 we had an easy delivery to beautiful 9 lb. 5.9oz Quinnlin Bay Roberts. She had big eyes, amazing skin and long fingers and toes. She joined proud brother Julian Gage, a blond boy with an unbelievable smile.

It was noticed that Quinn had enlarged kidneys during a routine prenatal appointment at 36 weeks. Two days after her birth, we were told over the phone by an on-call pediatrician that she had an incurable kidney disease - she used the word "polycystic." We were instructed to see our pediatrician the following day and set up an appointment with a nephrologist as soon as possible. Twenty minutes later we were on our way home with the devastating news that our sweet baby girl would eventually be in kidney failure and might not survive. No renal function test was preformed to see what her kidney function was.

We saw a pediatrician (not our regular Doctor) the following day and once again we were given very bleak odds – "be grateful for the time you will have her" were the words we heard. The information we could find didn't leave us with much hope. Finally, a nephrologist ordered tests for renal function and that same week we met with him where we learned Quinn's prognosis – an eventual kidney transplant early in her life, as well as complications with her liver, high blood pressure among other things. For the time being, her kidney function is considered "normal."

Soon came what almost all parents of children with PKD know – monitoring of blood pressure and kidney function. Quinn started medication for high blood pressure at two weeks old. Three months later we had our 3-year-old son tested and we learned that he too has ARPKD – he has "normal" kidney function as well. Medications for both, weekly trips to the Doctor for blood pressure monitoring, and monthly blood tests. To complicate matters, Gage and Quinn also have a rare eye disorder (Ocular Motor Apraxia) causing developmental delays. Quinn has begun physical and occupational therapy and Gage has been in therapy for a year and a half.

This is not the journey we expected, but it is where we must go. We continue to have hope for Gage and Quinn. We have to.

Choosing a Transplant Center

By Lori S. Spier

ARPKD will inevitably lead to transplant. Choosing a transplant center is one decision among many that we must make on the road to transplant.

Our family has started to investigate the options available. We hope that by sharing our information with all of you that we will all make better decisions. I also encourage you to write back to the newsletter with anything you found to be different in your exp erience.

Proximity

In general, you need to balance several important factors such as the proximity of the transplant center to your home and family support system. In the best case scenario, the hospital stay is on average one to two weeks. However, you will need to make frequent trips back to the center for blood tests and examinations. There are also cases where the hospital stay can be much longer depending on the complications involved. If you are too far away from home it can make running your life exceptionally difficult in terms of working and managing your other children. It is also important to have a support system nearby, as the psychological aspects of going through this process can be very stressful.

Survival Statistics

Another important factor to consider is the survival rates of the center, along with the volume of transplants done and the experience of the surgeons and the center staff as a whole. The United Network for Organ Sharing can provide this information via the internet at www.unos.org. or by calling 888-TXINFO1. Be mindful however, that these statistics do not take into account variables such as how sick the patient was prior to transplant, or how difficult the surgery was. Additionally, many centers reject difficult cases while others do not turn anyone away. Some inner city transplant centers may not do as well statistically, due to uneducated parents who are not as attentive to compliance with the medication or spotting signs of rejection. Therefore, statistics can be misleading and it is always smart to discuss your choices with a doctor that you trust. Also, speaking with other parents who have already been through the process is a great way to learn from their experiences.

Experience

Programs that are in existence for many years are usually more prefe rable to those in existence only a few years. Also keep in mind your long term needs. Many ARPKD patients need both a kidney and liver transplant, and you want to make sure that your center is experienced at both.

Follow-Up

Furthermore, our doctor advised us that some programs have surgeons follow post-transplant, while others have nephrologists follow the patients. He strongly felt that the nephrologist should follow patients as they are better acquainted with blood pressure and electrolyte issues.

Additional Resources

The above information is by no means exhaustive and I encourage you to obtain information from UNOS and from the following organizations:

1. American Association of Kidney Patients, 100 S. Ashley Drive, Ste.280, Tampa FL,33602. 800 749-2257. http://cybermart.com/aakpaz/aakp.html
2. American Liver Foundation,1425 Pompton Avenue, Cedar Grove NJ 07009,800223-0179, www.liverfoundation.org
3. National Kidney Foundation,30 East 33rd Street, New York, NY 10016, 800 662-9010, www.kidney.org
4. Organ Transplant Support Inc., PO Box 471, Naperville, IL 60566-0471,630 527-8640, www.inil.com/users/matt/ots-1htm
   Transplant Recipients International Organization, 1000 16th Street NW, Washington DC 20036,800 874-6386, www.primenet.com/~trio
5. Stanford Team Prevents Kidney Transplant Rejection Without Drugs @ http://www-med.stanford.edu/school/

DIAGNOSIS, PROGNOSIS & FINDINGS in ARPKD & CHF

It is now well established that terms such as Infantile Polycystic Kidney Disease (PKD), Early Childhood PKD, Late Childhood PKD, Adolescence PKD, Juvenile PKD, Early Onset PKD and Adult PKD are antiquated and obsolete; either a person has Autosomal Recessive Polycystic Kidney Disease (ARPKD) or Autosomal Dominant Polycystic Kidney Disease (ADPKD) or some other form of cystic kidney disease.

Originally, the ARPKD diagnosis included the person's age at time of discovery. For example, if ARPKD was found during infancy, then the label Infantile PKD was given. However, if that same child was not discovered to have ARPKD until adolescence, then an Adolescent PKD label was given.

Ultrasound may begin to visualize large echogenic kidneys, also described as "bright", from 13 weeks gestation, with low or absence of amniotic fluid (oligohydramnios) beginning after 20 weeks gestation. There have been cases where ARPKD was not visualized on sonogram until the 3rd trimester or after birth. A prenatal finding may also include absence of fetal bladder filling. Huge flank areas may complicate vaginal delivery. For those severely affected, Potter' facies or Potter's syndrome may be present, seen as deep set eyes, broad, flattened nose, abnormally small jaw bones, and low set ears; additionally, deformities of the extremities due to fetal restriction and compression caused by insufficient amniotic fluid may occur. It was reported in the "American Journal of Medical Genetics", Jan 2000 issue, that the "Syndrome of Autosomal Recessive Polycystic kidneys with skeletal and facial anomalies is not linked to the ARPKD gene locus on chromosome 6p".

Kidneys can be grossly enlarged, their size peaking at 1 to 2 years of age, with stabilization at 4 to 5 years of age. With time, sonographic patterns change due to the progression of cysts and fibrosis, changing the typical kidney shape. Older patients may be mistakenly identified as having ADPKD. Contrast-enhanced computerized tomography (CT) has been effective in visualizing the collecting tubules, which are dilated in ARPKD. Ultrasounds have replaced intravenous pyelogram (IVP). Glomerular Filtration Rate (GFR) may improve during the first 6 months of life from kidney maturation. One source stated that a third of the ARPKD population would need dialysis or kidney transplantation by 10 years of age. However, renal decline occurs with great variability in age, with some cases of end stage renal failure (ESRF) not occurring until early adulthood.

Amniocentesis and direct genetic testing cannot yet offer an ARPKD diagnosis. Prenatal testing is available for families who have had one confirmed ARPKD birth. This is an indirect diagnostic technique, based on gene linkage and the PKHD1 locus, and dependant on DNA from current or previously affected children and their parents. Diagnosis should be differentiated from other kidney disorders, including dysplastic kidneys, glomerulocystic kidney disease, diffuse cystic dysplasia or medullary sponge kidney.

Prognosis, especially for those who survive the newborn period is far less bleak then once thought. Infants with ARPKD who survive mechanical ventilation have a good chance of survival. The 5-year survival rate is 80 to 95% for those who survive the neonatal period (pertaining to the first 4 weeks after birth). Because of improved ESRD (End Stage Renal Disease) treatment, and more effective control of both systemic and portal hypertension, survival into adulthood is common.

Correlation of disease symptomatology with age at time of diagnosis has been suggested. It was thought that ARPKD individuals who were diagnosed early in life exhibited more kidney manifestations, with little, or sometimes no apparent liver involvement, while those diagnosed later in life, up to adolescence, (seldom into adulthood), manifested more clinical liver complications with little or no kidney manifestations. This description does not however take into account the prominent liver manifestations that occur in children with early kidney transplants who initially exhibited no liver problems.

Because of continued improvements in mechanical ventilation, neonatal support, control of hypertension, management of ESRD and transplantation, the ARPKD population is now living well into adulthood.

More information can be found at www.arpkd.org

Health Insurance

In this day of managed health care, costs sometimes take precedence over care, decisions sometimes are made accordingly to costs. An informed patient (and determined) often gets the best-managed care. If an HMO says no to something you feel is needed, you can appeal their decision. A limited number of appeals may apply, check with your insurance policy.

RESOURCES:
CHIP (Children's health Insurance Program)- Is a U.S. federal law to use state and federal dollars to provide health insurance for uninsured children. Also known as Title XXI and SCHIP. States create their own CHIP plans and send them to the federal government for approval.

FAMILY VOICES-A grassroots effort speaking on behalf of children with special health care needs, extensive information including insurance assistance @ 1-888-835-5669 or www.familyvoices.org

HIPAA-Health Insurance Portability and Accountability Act- @ http://www.hcfa.gov/medicaid/HIPAA/default.asp

Healthinsuranceinfo.net!-Consumer Guides for Getting and Keeping Health @ Insurance: http://www.healthinsuranceinfo.net/

Thoughts on Living with ARPKD and CHF

"Every situation is so different, yet so much alike", by Diane Nelson, mother of 28 year old living with ARPKD and CHF.

Most if not all ARPKD parents are completely unprepared for the diagnosis. Initial grief can be overwhelming. Diagnosis is frequently followed by a quest to understand the disease, (often resulting in minimal and inconsistent information). There may be shock and disbelief among family and friends. For those who survive the newborn period, it is difficult to understand and accept the invisible abnormalities lurking inside our infants and children, they look so healthy and normal. As we slowly accept the diagnosis and circumstances, we may experience roller coaster emotions- and cope by using denial, sadness, humor or anger. Recognize these are natural responses to grief, and that men often grieve differently then women. We may harbor fear and fear the unknown future. To help ease the fear and isolation, alleviate powerlessness and hopelessness, educate yourself. **Education may be your key to coping**. Knowledge can be powerful, it diminishes fear, and an informed parent is a prepared parent. Increase your sense of control; find out everything you can about your child's disease. Develop a strong family-professional team, a mutual commitment that recognizes and respects the knowledge, skills and experience that each brings to the relationship. Promote collaborative decision making with the medical team, because who knows your child better then you? Ask questions and be recognized as your child's expert. Medical professionals have a snapshot view, but you see your child on a daily basis. Parents are a valuable source of information. Become proactive to enhance outcomes and ensure quality care, you can make a tremendous difference.

Listserve for ARPKD

There is now a listserve for ADULTS living with ARPKD and CHF, established by the ARPKD/CHF Alliance. An exchange of information, networking, and support, comments, questions, and discussion related to ARPKD and CHF are welcome. To subscribe, go to: http://listserve.geneticalliance.org/mailman/listinfo/arpkdadults

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Letter From Slovenia—

From: Veronika Poje

In 1996, I gave a birth to healthy baby girl. I named her Sara. When she was 3, we left her father because he become violent. I met Robert and soon we decided to have a baby. I got pregnant and everything went well. Even at ultrasound they said everything is ok. Our family never had problems with kidneys. So on June 13th this year Hana came. Labour lasted only 20 minutes. Everything went well, and then the nurse said that Hana has bigger stomach and she took her to ultrasound. Some 20 minutes later, 3 doctors came in room and they started to talk about some problems with kidneys. They took Hana to intensive care and I went to my room. I cried all night because I didn't know what's wrong. At 10 AM next morning, I went to see Hana and they said her blood test was very bad. The same day I got her in my room, but no one told me, what's wrong with her. In our country it is a law, that doctor doesn't necessary tell all things about diseases - just as they think it wise. So they told me that maybe her kidneys will not work well, but that they know a boy who is 4 and he's doing fine. After 9 days in hospital we came home. Her blood tests were much better and we had no medicine. First thing I did when I came home, was go to Internet to see about her disease. When I started to read, I couldn't believe my eyes. As I saw, we are lucky, Hanna is alive. Second shock was disease has no treatment. I just cried and I didn't know what to think. I called Robert and told him. We were down, sad. I was afraid, I suddenly didn't' t know how to be with Hana. I cried all nights and lost almost all my milk. Thank God, Sara was on holidays at that time. After 7 days we had to go to hospital for checking. Her blood pressure was high - docs had to find right medicine. Her Natrium (Na) was low - 127. I was again down, cause I couldn't stay with Hana in hospital over night. Our children hospital is old and small - they have 8 beds for breast feeding mothers and because I live just 30 kilometers away, they didn't give me the bed. I woke up every day at 6:00 AM, went to hospital, sat beside Hana all day till 11:00 PM and went home. During night I pumped milk from breasts and took it to hospital for Hana in the morning. Doctors took tests -- her kidneys work left 46% and right 56%. Ultrasound showed cysts on livers too. After one month in hospital she was released, because growing of cysts stopped 3 weeks ago and we got medicines. We were twice on cheks and everything was good, just Natrium is always low - even if she drinks so much Na water. In those 2 moths, I changed a lot. It was hard to cope with thought that my child will have disease whole life. I've never been to hospital as child and actually I was afraid of hospitals. But reading and talking about ARPKD makes things easier. I lost fear in front of Hana although I start to panic if I am not sure why she is crying. A few days after coming home from hospital I felt like I am not able to take care of Hana because there is no nurse or doctor around. But now I am strong and I got my self esteem back.

It was hard to hear Hana's crying when they took her blood and I couldn't do anything. It was sad looking at baby whose arms were all blue from needles. I feel sadness because I can't help or protect my child. I somehow got stronger and Robert helps me a lot and there is also Sara who adores Hana.
And last week I spoke to a doctor who is the best for ARPKD in Slovenia. I told him that I joined to parents of ARPKD children in States. He told me that he has some children with ARPKD, but there is no such group in our country. So we decided to found it. Doctor said he has problems to talk with parents - how to tell them and how much to tell them. When he saw, how much I know about disease, he talked with me openly. I can't say that I am not scared for the future, but we try to live as normal a life as it's possible. We enjoy every day, every moment. Like one leader said: "Live like there will be peace 100 years, and be prepared that there will be war tomorrow."

At the end I would like to thank my friend Gloria, who helped me to do first steps and give me courage to cope with disease and become strong person!

Veronika Poje
Slovenia, Europe
Written in 2001

(Note: If you would like to correspond with Veronika, please send her an e-mail at robert.kriz@siol.net. She would love to hear from you!)

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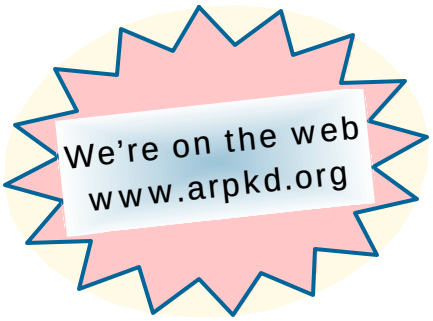

In February, 2002 we launched a new web site specific to ARPKD/CHF. The site can be found at www.arpkd.org. We invite all families and friends to view this site for updates and information regarding research and how to make a contribution to our cause. Since this is a work in progress, we welcome any comments you may have … we are all in this together. Thank you!

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If you would like to share your story/experience with our ARPKD/CHF families to be published in our next newsletter, please send to Gloria Dengler-Mondaruli at: miglo@pipeline.com