Autosomal recessive polycystic kidney disease (ARPKD) is a relatively rare form of PKD, affecting approximately one in 20,000 children. ARPKD often causes death in the first month of life. For the 70% of ARPKD children who survive the newborn period, approximately one-third will need dialysis or transplantation by age 10.

Previously thought to be a fatal condition, the prognosis for children with ARPKD has improved dramatically. Twenty years ago, only half of the children born with the disease survived to their 10th birthday, but now that percentage has increased to 85%.

Cost of ARPKD

There is no treatment or cure for ARPKD. Dialysis and transplantation are the only options to treat the damage the disease has caused. Dialysis costs nearly $100,000 annually and a kidney transplant costs $250,000. With the damage beginning so young, ARPKD patients may need multiple transplants throughout their life. Children with ARPKD often have congenital hepatic fibrosis, which can result in the need for a liver transplant, which is more than $500,000.

Diagnosis

Typically in ARPKD, the kidneys appear to be larger than normal. In some babies, prenatal ultrasound can detect the enlarged kidneys as early as 18 weeks after conception. Prenatal genetic testing is possible.

Symptoms of ARPKD

Prenatally

- Diminished amniotic fluid levels during pregnancy
- Enlarged kidneys on fetal ultrasound
- Lung immaturity and functioning issues

Immediately after birth

- Enlarged kidneys due to cysts
- Breathing problems due to lack of space because of enlarged kidneys and decreased urine production.
- Ventilation is frequently required to sustain life
- Excessive urine production
- Hypertension
- Growth problems
- Congenital hepatic fibrosis
- Enlarged spleen with low red blood cell, white blood cell and platelet counts

Health management in ARPKD

Families should work with specialists in several areas, including liver, kidney and lung specialists, to monitor the child’s health. The numerous appointments result in missed school and work, which is a huge burden and disruption to normal life.
PKD Foundation support
The PKD Foundation is the only organization in the U.S. solely dedicated to finding treatments and a cure for PKD to improve the lives of those it affects. We do this through promoting programs of research, education, advocacy, support and awareness on a national level, along with direct services to local communities across the country. Our vision is that one day no one will suffer the full effects of PKD. Visit pkdcure.org to learn more.

Amanda Jaska – San Antonio
At 18 weeks into my second pregnancy, we went in for a check up. The technician’s face just dropped as he looked at the ultrasound images. The next few minutes turned into weeks and months of anguish and stress. My doctor had tears running down her face. She told me my baby had ARPKD and his kidneys were filled with cysts. She said these babies usually die when they’re born. I couldn’t believe what I was hearing. What should have been a time of joyful anticipation and preparation became a torturous wait. It was such an emotional time, talking to priests and planning memorials to prepare for the worst.

Miraculously, Gus was born at 37 and a half weeks by C-section because his stomach was too distended for a traditional birth. But other than his big belly, he looked perfect. They took him to the NICU, but he didn’t need any feeding tubes or a ventilator.

Living with ARPKD has been a difficult adjustment for the whole family. His kidneys have increased in size about 1 cm each, but his function is still great. We did find out this year he also has Caroli’s disease, a rare liver disease that can happen in children with ARPKD. So we also have to keep a close eye on his liver function and watch for infection.

He is a happy, three-year-old boy that loves tractors and football. We are grateful for every day with Gus.

The Foundation has invested more than $2 million since 2006 for research, support, education and awareness for ARPKD and ADPKD in children and congenital hepatic fibrosis (CHF), a disease closely associated with ARPKD.

PKD Parents Chapter
The PKD Parents Chapter provides support for parents of children with ARPKD as well as ADPKD. This Chapter has served more than 600 families since 2000. Once a family with a child living with PKD reaches out to the Foundation, a volunteer from the PKD Parents Chapter contacts them for support, compassion and guidance. Families can be connected with experts and other local families and are supported in many ways. To learn more, email pkdparents@pkdcure.org.