



ARPKD PATIENT HANDBOOK

Understanding and living with autosomal
recessive polycystic kidney disease and
congenital hepatic fibrosis



PKD FOUNDATION
Polycystic Kidney Disease



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Introduction

The purpose of this handbook is to provide information about **autosomal recessive polycystic kidney disease (ARPKD)** and **congenital hepatic fibrosis (CHF)**. It will be useful to children and families who have been diagnosed with ARPKD/CHF, as well as family members, caregivers and health professionals. It is not intended for those affected by autosomal dominant polycystic kidney disease (ADPKD).



What is ARPKD?

ARPKD is a rare genetic disease that occurs in approximately 1 in 20,000 children. It affects all individuals equally, regardless of gender, race or ethnicity.

Many children with ARPKD are diagnosed before birth if prenatal ultrasound shows **enlarged, bright-appearing “echogenic” kidneys** and **low amniotic fluid** levels.

Some babies can have **poor kidney function** and underdeveloped lungs at birth, and some die in the first month of life from these complications. If a child with ARPKD survives the newborn period, the chances of survival are good. For these children, approximately one-third (1 in 3) will need dialysis or a kidney transplant by 10 years of age.

Some children are diagnosed with ARPKD later in childhood. Children who are diagnosed later generally have milder disease progression, and approximately one-quarter (1 in 4) of them will need dialysis or a kidney transplant by 30 years of age.



What is CHF?

All children with ARPKD have the liver abnormality Congenital Hepatic Fibrosis (CHF), but it does not always cause symptoms or problems. CHF can cause the liver to be larger than normal.

Children with CHF have **dilated bile ducts** within the liver, which are at risk of getting an infection called **cholangitis**. CHF can also slow down blood flow from the intestines and spleen to the liver, which is called **portal hypertension**. This causes high pressure in the veins around the esophagus, stomach, and intestines, and the veins can become dilated. These dilated veins, called **varices**, can rupture and cause potentially life-threatening gastrointestinal bleeding.

Portal hypertension can also cause the spleen to become enlarged and break down blood cells, which can cause low white blood cell counts, anemia, and low platelet counts. About 5-10% of children (1 in 10-20) with ARPKD/CHF will need a liver transplant.

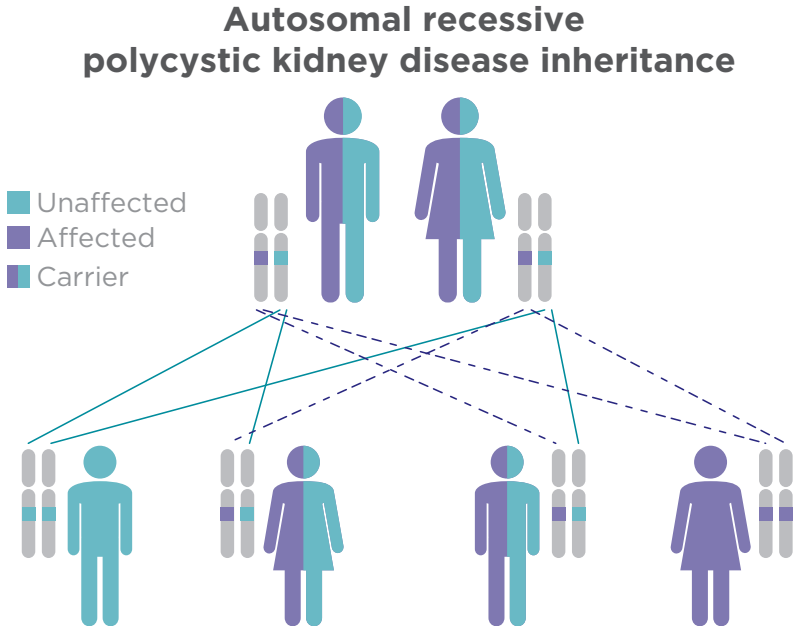
Genetics of ARPKD

Inheritance

ARPKD is a genetic disease that is caused by a **mutation** (mistake) in a gene called **PKHD1** that is important for proper development of the kidneys and liver. ARPKD is inherited in a **recessive** manner – this means that a child must inherit **two** mutated **PKHD1** genes (one from each parent) to be affected with ARPKD. A person with one normal and one mutated **PKHD1** gene will be a **carrier**, and will not have any symptoms of ARPKD.

When both parents are carriers of an abnormal **PKHD1** gene, each child has:

- A 25% (1 in 4) chance of being affected by ARPKD
- A 50% (1 in 2) chance of being an unaffected carrier
- A 25% (1 in 4) chance of being unaffected and not being a carrier



Most people do not know that they are carriers of ARPKD until they have a child affected by ARPKD. There are many different types of *PKHD1* mutations. The symptoms of ARPKD do not always correlate with the type of mutation, and sometimes, siblings with the same mutations can have very different ARPKD symptoms.

Genetic testing

It is possible to test for ARPKD using **genetic testing** to sequence the *PKHD1* gene to look for mutations. Testing will typically find a mutation in about 85% of affected patients. There are some other childhood kidney diseases that can have similar symptoms to ARPKD. Some laboratories can test for multiple genetic kidney diseases at the same time, which can be helpful in situations where the diagnosis is not clear. In a child with a suspected diagnosis of ARPKD, testing is done using a blood sample. If genetic testing is desired before birth, a procedure such as **chorionic villus sampling (CVS)** or **amniocentesis** is needed to get a sample of the fetus's genetic material. It is also possible to perform **pre-implantation genetic diagnosis (PGD)**, which is discussed on page 14.

Diagnosis and treatment

In utero: Before birth

Diagnostic tests



Prenatal ultrasound

ARPKD is often first suspected based on routine prenatal **ultrasound**. The ultrasound may show symmetrically **enlarged kidneys** that appear brighter than normal “**echogenic**” because they contain many tiny fluid-filled cysts. Sometimes **amniotic fluid levels can be low**, because the fetus’s kidneys are not working properly and are not making enough urine. Sometimes, the **lungs appear underdeveloped**, due to compression from very large kidneys and/or from low amniotic fluid levels.

As mentioned before, a few other diseases can resemble ARPKD on prenatal ultrasound. It is therefore not always possible to make a definite diagnosis of ARPKD based on ultrasound, unless previous children in the family have a confirmed diagnosis of ARPKD. Also, a normal prenatal ultrasound does not always exclude a diagnosis of ARPKD, because sometimes kidney abnormalities may not develop until late in pregnancy or after birth.

Prenatal genetic testing

Since ultrasound does not always detect abnormalities until later in a pregnancy, genetic testing can be helpful to make an earlier diagnosis of ARPKD if the parents are known to be carriers of ARPKD. As mentioned before, a procedure such as CVS or amniocentesis is needed to get a sample of the fetus’s genetic material for testing. CVS can be done as early as 10-12 weeks of pregnancy, and amniocentesis can be done after 15 weeks of pregnancy. Early genetic testing can be helpful for some families who are deciding whether to continue a pregnancy, particularly if they have had a previous child severely affected with ARPKD.

If typical findings of ARPKD are discovered on a prenatal ultrasound, genetic testing is usually not necessary to confirm the diagnosis.

After diagnosis: What are the next steps?

Prognosis

It is often very difficult for doctors to predict how severely a baby with ARPKD will be affected based on prenatal ultrasound or genetic testing. Sometimes, there are such severe signs on a prenatal ultrasound (for example, severely underdeveloped lungs or lack of amniotic fluid) that the chances of the baby surviving after birth may be very small. However, there are also times when a baby who did not appear to have severe abnormalities on prenatal ultrasound goes on to develop severe, life-threatening complications after birth. On the other hand, there are also times when a baby who appeared to have severe abnormalities on a prenatal ultrasound is able to survive after birth. Genetic testing also does not always help in predicting a baby's outcome, because the type of *PKHD1* mutation does not necessarily correlate with symptoms.

Finding guidance

It can be overwhelming for families to receive an ARPKD diagnosis before birth, and they can be faced with many difficult decisions. It is often helpful to seek guidance from doctors at specialized medical centers who have experience taking care of families affected by ARPKD. Doctors can include obstetricians (OBs) (particularly those with advanced training in maternal-fetal medicine), pediatricians, pediatric nephrologists (kidney doctors), and neonatologists (pediatricians specializing in newborn care). Hospital social workers, psychologists, and child life specialists can be a good source of support. Advocacy groups, family, and friends can also be good sources of support. The PKD Foundation's PKD Parents Chapter can connect you with other families who have similar experiences.

In some cases, if the chance of survival after birth is thought to be extremely small, families may choose to terminate the pregnancy. If the family chooses to continue the pregnancy, it is important for the mother to be followed closely throughout her pregnancy by an OB with experience in taking care of women with medically complex pregnancies.

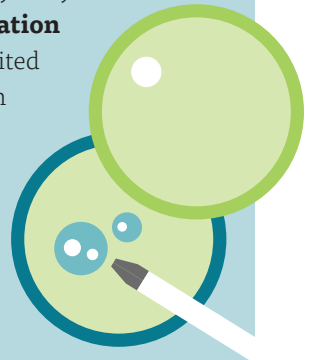
Making care plans

It is also important for the family to think about a care plan for the baby's birth, including choosing the hospital where to deliver the baby. If the family wishes to pursue maximum medical support, it is important to choose a hospital with an advanced neonatal intensive care unit (NICU) and specialists with experience taking care of babies with severe diseases such as ARPKD, including neonatologists, pediatric nephrologists, and pediatric surgeons. The family may also consider asking about the hospital's experience with performing dialysis in infants. In rare cases, medical professionals do not recommend or support offering medical care after birth if the survival outcome seems poor. It is important to remember this is a very personal choice, and it may mean changing doctors or centers to find a care team that fully supports the family's wishes.

Pre-implantation Genetic Diagnosis (PGD)

In families with a previous child severely affected by ARPKD, PGD can be a valuable way to prevent another child from being affected with ARPKD. To carry out PGD, the parents must have genetic testing to find out which *PKHD1* mutations they carry.

Then, a couple must undergo **in vitro fertilization (IVF)**, meaning that the sperm and egg are united in a laboratory. The resulting embryos can then be tested for *PKHD1* mutations, and only the embryos without ARPKD are transferred into the mother. Couples considering PGD will need to seek care at a medical center with the required expertise, including fertility experts and genetic counselors.

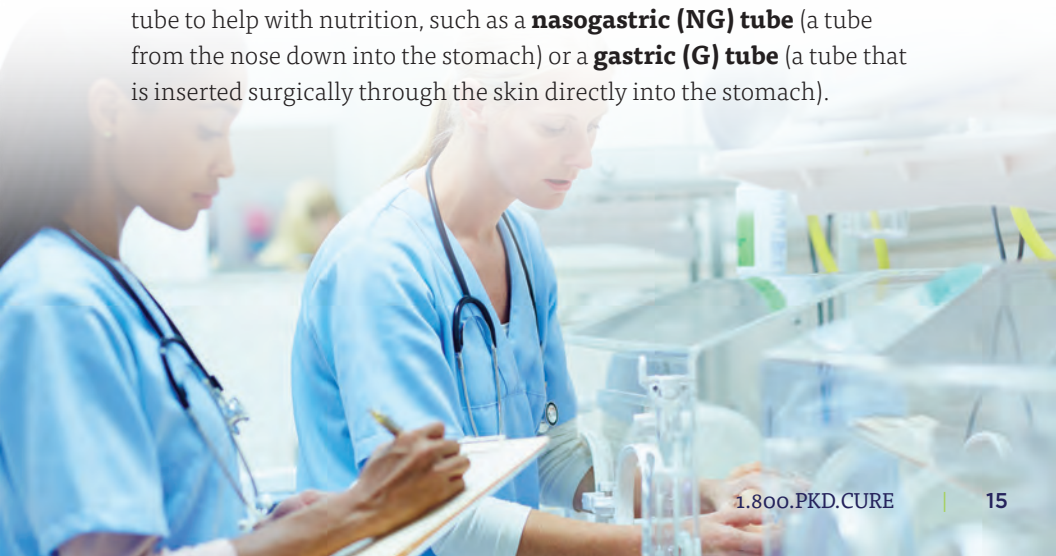


Immediately after birth

At birth, many babies with ARPKD have **breathing problems due to underdeveloped lungs**. Many babies will require special support such as mechanical ventilation (breathing machine), or may develop a pneumothorax (collapsed lung).

Very enlarged kidneys can also cause breathing problems due to compression of the lungs, and they can also make it difficult for babies to eat due to compression of the stomach and intestines. Sometimes, doctors may recommend that one or both kidneys be surgically removed to make more room in the baby's abdomen. However, the possible benefits of removing the kidneys needs to be balanced with the risks of surgery and the earlier need for dialysis if the kidneys are removed. Dialysis is discussed on pages 20-21. Every baby's care is unique, and these options need to be carefully considered and discussed for each child individually.

Because of the complications described above, babies with ARPKD can have **difficulty feeding** by mouth. Many babies will require a feeding tube to help with nutrition, such as a **nasogastric (NG) tube** (a tube from the nose down into the stomach) or a **gastric (G) tube** (a tube that is inserted surgically through the skin directly into the stomach).



Losing a baby with ARPKD

Unfortunately, even with advanced medical care, about 1 in 4 babies who are diagnosed with ARPKD before birth die in the newborn period, most often due to breathing complications. Families and doctors may need to work together to make difficult decisions about how aggressively to pursue medical interventions, or decide to withdraw medical care. It is important for families to get support from other family members, friends, and medical professionals such as palliative care providers to help them navigate these difficult decisions.

Preserving tissue for genetic testing

When a baby dies, parents may request that tissue be saved for further analysis, donated to ARPKD research, or both. In order to provide geneticists with the information they need to counsel for future pregnancies, they must have genetic material, which is usually comprised of kidney tissue and blood samples. It is ideal to discuss and plan for this possibility, because it is often difficult to make these decisions when one is newly grieving. The Coordinators of the PKD Parents Chapter can provide information and guidance through this process.

Bereavement photography

Among the difficult discussions around care plans for a baby with severe ARPKD is the option of bereavement photography. In the event of a severe diagnosis, a professional photographer trained to be sensitive in such circumstances can offer their services to capture images of the baby and family. Some families have reported that tastefully done photos at the end of life, or after life, have added in their healing process and are immense treasures. In some cases, the photographer may want to speak with someone on your behalf about the diagnosis and potential outcomes. Many centers with NICUs have experience in working with photographers. See Resources on page 58.

Kidney function, dialysis, and transplantation

The kidneys of individuals with ARPKD tend to be very large and are filled with multiple tiny cysts. The cysts arise from small tubules in the kidneys, called **collecting ducts**, which have become abnormally dilated. The multiple cysts cause the kidneys to appear echogenic on a kidney ultrasound. The cysts make it difficult for the kidneys to work properly, and most individuals with ARPKD will develop **chronic kidney disease (CKD)**. This means that the kidneys are not functioning properly and cannot properly get rid of the body's waste products. In most people with ARPKD, kidney function becomes worse over time, and they eventually develop kidney failure, called **end-stage renal disease (ESRD)**. When ESRD develops, treatments to replace kidney function such as **dialysis** or a **kidney transplant** are needed.

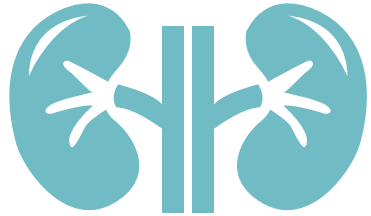
The age at which individuals with ARPKD develop ESRD varies widely from person to person. In general, individuals who were diagnosed with ARPKD as newborns tend to develop ESRD at an earlier age. Among children who are diagnosed with ARPKD at or before birth and who survive the newborn period, about one-third (1 in 3) will develop ESRD by the age of 10. Among children who are diagnosed with ARPKD after the first year of life, about one-quarter (1 in 4) will develop ESRD by the age of 30.

Chronic kidney disease (CKD)

CKD means that an individual's kidneys are not fully able to perform all of their normal functions. CKD can be caused by many different diseases, including ARPKD.

The main functions of the kidneys include:

- Getting rid of **waste products** from the body
- Controlling the body's **fluid balance**
- Regulating levels of the body's **electrolytes** (chemicals such as sodium, potassium, and bicarbonate)
- Regulating **blood pressure**
- Producing a hormone called **erythropoietin (EPO)** that tells the body to make red blood cells
- Activating **vitamin D**, which is important for bone health



There are five stages of CKD, based on the level of kidney function.

Kidney function is measured using the **glomerular filtration rate (GFR)**, which is calculated based on blood levels of creatinine.

Creatinine is a normal waste product that is made by the muscles, then is filtered by the kidneys and excreted into the urine. If kidney function is abnormal, the body cannot get rid of creatinine as efficiently, and the blood creatinine level gets higher. A higher creatinine level means worse kidney function.

Symptoms of CKD in children can include tiredness, fluid retention, poor appetite, poor growth, problems with bone health, abnormal blood levels of electrolytes, anemia (low red blood cell counts), high blood pressure, and learning/developmental problems.

In the early stages, many of the symptoms and complications of CKD can be managed with medications. These can include medications to manage electrolyte levels, blood pressure medications, vitamin D and iron supplements, EPO injections, and growth hormone injections. Dietary changes may also be needed. However, as CKD progresses and an individual develops Stage 5 CKD (ESRD), treatments to replace kidney function such as **dialysis** or a **kidney transplant** are needed.

CKD stages

- 1 Stage 1:** Includes signs of mild kidney disease, with a normal GFR showing 90% or higher kidney function.
- 2 Stage 2:** Includes signs of mild kidney disease with a GFR showing 60-89% kidney function.
- 3 Stage 3:** Includes signs of moderate kidney disease and a GFR showing 30-59% kidney function.
- 4 Stage 4:** Includes signs of severe kidney disease and GFR showing 15-29% kidney function.
- 5 Stage 5:** Includes signs of severe kidney disease and kidney failure (ESRD), with a GFR showing less than 15% kidney function.

Dialysis

Dialysis is a procedure that is used to replace some functions of the kidneys after they fail. This includes removing waste products and extra water, and balancing the level of different chemicals in the body such as potassium and sodium. There are two main types of dialysis: **hemodialysis (HD)** and **peritoneal dialysis (PD)**. Some babies need dialysis early in life if they are born with poor kidney function, or if one or both kidneys required surgical removal. Other children need dialysis later in life. The choice of dialysis type for an individual depends on many factors, including age and other medical conditions as well as family situation. In babies and young children, PD is generally preferred.

Hemodialysis (HD)

HD, or blood dialysis, uses a machine that acts as an artificial kidney to remove waste products and extra water from the blood and to balance chemical levels in the body. To perform HD, **access** is needed to get blood from the patient to the HD machine. In young children, the most common type of access is a catheter that is inserted into one of the large veins in the neck or upper chest. Access can also be created in an arm or leg by doing a surgery to connect an artery to a vein to make a bigger blood vessel (called a fistula or graft). HD is generally performed in a hospital or clinic. The amount of time needed for HD treatment can differ from one person to another, but in general each HD treatment lasts about 3-4 hours and is done 3-4 days per week.



Peritoneal dialysis (PD)

In peritoneal dialysis (PD), a special fluid called dialysate is put into the abdomen (belly) to remove waste products and extra water, and to balance chemical levels in the body. To perform PD, a plastic tube called a **PD catheter** is surgically inserted into the peritoneal cavity, which is the area within the abdomen that surrounds the internal organs. Dialysate fluid is put into the PD catheter. As the fluid sits in the peritoneal cavity, it absorbs waste products and extra water and corrects chemical imbalances. The used fluid is then drained out, and clean fluid is then put back in. Each time fluid is put in and drained is called an exchange, or cycle. Multiple cycles are usually needed each day, which are usually done using an automatic machine (called a cycler) overnight while the person is sleeping. Long-term PD is usually done at home by the patient and/or family, and requires less frequent visits to the clinic or hospital compared to HD.



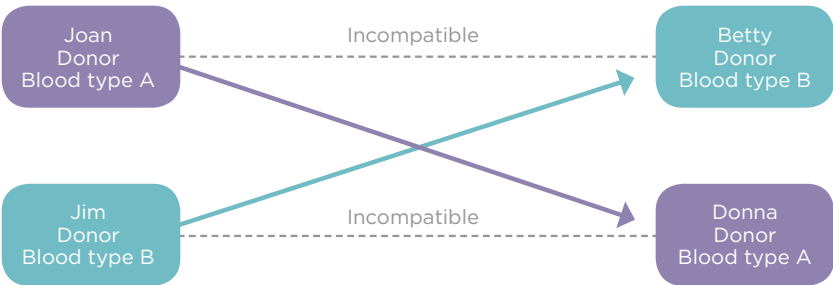
Complications of dialysis

Both HD and PD can be associated with complications. Children undergoing dialysis should be cared for in specialized centers with experience in pediatric dialysis, so that possible complications can be closely monitored and treated. HD and PD catheters require meticulous care to prevent them from becoming infected. HD and PD catheters can sometimes stop functioning due to blockage or changes in positioning, and may require replacement. The changes in body chemical and water balance in children undergoing dialysis can sometimes cause symptoms such as fatigue, changes in blood pressure, weight gain or loss, cramping, or dizziness. Children on dialysis require close medical follow-up to monitor and prevent these problems.

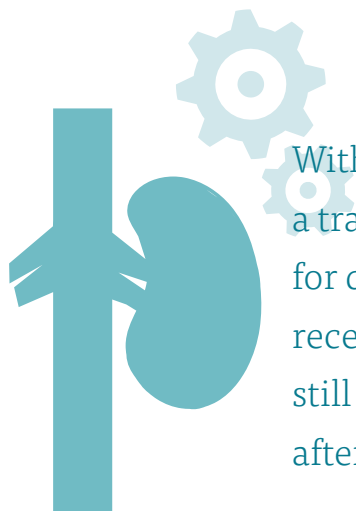
Kidney transplant

A kidney transplant is done by taking a kidney from a donor and surgically placing it into the recipient. The donor can be a **living donor** (for example, a parent, other relative, or an unrelated person), or a **deceased donor** from a national registry. **Paired donation** (see figure below) involves two or more pairs of living kidney donors and transplant candidates who do not have matching blood types. The candidates “trade” donors so that each candidate receives a kidney from a donor with a compatible blood type. This type of exchange often involves multiple living kidney donor/transplant candidate pairs and can join incompatible pairs from different centers or even different parts of the country.

Paired Donation



Kidney transplants must be performed at specialized medical centers with transplant programs. Before transplant, many tests are done to make sure the donor’s kidney will be compatible with the recipient. Living donors undergo extensive testing to make sure they are healthy enough to donate a kidney.



With good medical care, a transplanted kidney can last for decades: 1 in 2 children who receive a kidney transplant will still have a functioning transplant after 20 years.

Some children with ARPKD will need one or both kidneys to be surgically removed in order to make room for the transplant kidney in their abdomen. After a kidney transplant, medications must be taken for life to prevent rejection of the kidney. With good medical care, a transplanted kidney can last for decades: 1 in 2 children who receive a kidney transplant will still have a functioning transplant after 20 years. However, most people who receive a kidney transplant in childhood will eventually need one or more repeat kidney transplants or may also need dialysis while waiting for another transplant.

Some children who have severe kidney and liver involvement from ARPKD may require both kidney and liver transplants. These can either be done one after the other, or at the same time, depending on the individual patient's needs.

Common challenges for children with ARPKD



High blood pressure (hypertension)

Hypertension occurs in most (about 3 in 4) children with ARPKD. Blood pressure can be difficult to control, particularly in babies with ARPKD. It is common for babies and children with ARPKD to need multiple medications to control blood pressure. It is important to control the blood pressure properly to prevent complications such as kidney damage, heart disease and stroke.

Low sodium levels in the blood (hyponatremia)

Low blood sodium levels occur commonly in babies with ARPKD. Sometimes, babies with ARPKD are treated with diuretics (medicines to help the body excrete water) and/or sodium (salt) supplements.

Urine concentrating abnormalities

Some children with ARPKD produce large amounts of urine because their kidneys cannot adequately concentrate their urine. They may have higher fluid intake requirements than healthy children, and may have trouble with bedwetting or may need more frequent bathroom visits.



Urinary tract infections (UTI)

Children with ARPKD are at risk for UTIs, possibly due to slow urine flow in the cystic, dilated collecting ducts. About 1 in 3 children with ARPKD will develop a UTI, and these infections are more common in females. It is important to test for a UTI in any child with ARPKD who develops painful urination, cloudy or foul smelling urine, abdominal pain, and/or fever.



Feeding, growth and development

Many children with ARPKD can have difficulties with feeding and growth. This can be due to decreased food intake due to poor appetite and/or compression of the stomach and intestines by large kidneys, which can cause reflux or an early sensation of fullness. Many children with ARPKD need a special feeding tube such as an NG or G-tube to help them take in enough nutrition. Some children may also need treatment with growth hormone injections to help them grow.

Many kidney diseases, including ARPKD, can also affect a child's mental, social and educational functioning. Some of these problems are due to the fact that a child may miss school or other social and educational activities because of medical appointments or hospitalizations. Some problems are also due to the direct effects of kidney disease on brain functioning. It is important for children with ARPKD to be evaluated for learning problems and to receive appropriate support in school if needed.

Congenital Hepatic Fibrosis (CHF)

All children with ARPKD have a liver abnormality called CHF, because the same genetic defects responsible for kidney disease in ARPKD can cause liver disease as well. However, CHF does not always cause symptoms or problems. The extent of liver disease is difficult to predict, and some patients are more affected than others. Being aware of liver disease in ARPKD is important, as it can have a significant long-term health impact.

CHF can cause enlargement of the liver, dilation of bile ducts within the liver and slow blood flow from the intestines and spleen to the liver, which is called portal hypertension. This combination of problems is sometimes called **Caroli syndrome**.

In CHF, the liver tissue becomes tough and fibrous, scarred over time, causing the liver to change from being soft and supple to being more firm, inelastic and enlarged. In addition, the small bile ducts inside the liver can develop areas of dilation (cysts) and/or areas of narrowing (stenosis). Bile ducts drain the liver of bile and merge together into the common bile duct, which carries the bile to the intestine.

When it reaches the intestine, bile helps to digest fats and proteins, and to get rid of waste products or toxins. In CHF, the abnormal bile ducts may not drain bile properly, which can lead to poor bile flow. This can increase the risk for bile duct infections (**cholangitis**).

CHF causes a slowing of blood flow into the liver from the spleen and digestive organs like the esophagus, stomach and intestines. This can lead to increased pressure in those vessels, called **portal hypertension**. In patients with portal hypertension, high pressure in the veins around the esophagus, stomach and intestines can cause those veins to become dilated. These dilated veins, called **varices**, can rupture and cause potentially life-threatening gastrointestinal bleeding. Portal hypertension can also cause the spleen to become enlarged (**splenomegaly**).

Diagnosis of CHF

Signs and symptoms of CHF

Oftentimes, patients with CHF may not experience any symptoms of disease. As the liver becomes more fibrotic (scarred), or as bile duct cysts develop, symptoms could include fullness in the abdomen due to liver enlargement or fluid collection (called ascites). Sometimes, infections of the bile ducts, cysts or ascites can occur, which can cause fever and abdominal pain. Patients with portal hypertension can develop bleeding from ruptured varices, which can cause symptoms of bloody vomiting and/or bloody or dark stools. Any symptoms of bleeding in individual with ARPKD/CHF require urgent medical attention.

Patients with portal hypertension can have fullness in the abdomen due to enlargement of the spleen. An enlarged spleen also causes increased breakdown of blood cells, which can cause low white blood cell counts, anemia and low platelet counts. This can cause some patients to develop easy bruising.

Physical examination

Some signs of CHF can be detected by physical examination. These may include enlargement of the liver or spleen, or subtle changes in the skin or fingernails.

Laboratory testing

Blood tests can sometimes show mild abnormalities in liver function tests such as alanine aminotransferase (ALT), aspartate aminotransferase (AST), bilirubin (a component of bile), or gamma-glutamyl transferase (GGT). Some patients can have abnormalities in a blood clotting test called prothrombin time (PT). Blood clotting proteins are made in the liver and require vitamin K to be made. Some patients with liver disease cannot make enough clotting proteins because they cannot absorb vitamin K properly. Patients with portal hypertension and enlarged spleens can have low white blood cell counts, anemia, and low platelet counts.

Imaging

Ultrasound of the abdomen can show signs of liver fibrosis (such as abnormal liver texture or an enlarged liver) or signs of bile duct dilations or cysts. Ultrasound may also show enlargement of the spleen in patients with portal hypertension. Sometimes, magnetic resonance imaging (MRI) of the liver is performed to get a more detailed picture of the bile ducts.

Special procedures

If the diagnosis of CHF is in question, or if there is concern for significant fibrosis, a care team may want a liver biopsy to look at the liver tissue more closely. In general, these are relatively easy to perform, but may require a child to receive sedation. In brief, a small amount of numbing medicine is put on and into the skin outside the liver. Then, a small needle is inserted into the liver, and a “core” biopsy is obtained. There are possible complications of biopsies, including bleeding, so the decision to obtain one should include a risk and benefit analysis.

Treatment and monitoring of CHF

For patients with CHF, a daily multivitamin can be important for maintaining good nutrition. Having poor bile flow out of the liver can mean that less bile is available to help digest foods, especially fats. This may lead to a need for extra calories, and supplements of fat-soluble vitamins such as A, D, E and K. Patients with CHF should be careful to avoid other possible causes of liver damage. For example, a healthy diet with regular exercise is recommended to prevent fatty liver from developing. It is important to avoid alcohol and to make sure vaccines are up to date (to prevent contracting hepatitis B).

In children with dilated bile ducts, a medication called ursodiol is sometimes used to help bile flow in the liver. Children with abnormal bile ducts are also at risk of getting a serious infection called cholangitis, which causes fever and abdominal pain. It requires urgent treatment with antibiotics. Some children at high risk for cholangitis are prescribed a regular antibiotic to prevent infections. Children with ARPKD who have had a kidney transplant and are taking anti-rejection medications to suppress their immune systems are at higher risk for cholangitis. Therefore, special attention needs to be paid to symptoms such as fever and abdominal pain in these children.

Children with portal hypertension need to be monitored closely for symptoms of bleeding due to varices. Any signs of bleeding require urgent medical attention. Sometimes, a procedure such as **endoscopy**, also known as esophagogastroduodenoscopy or EGD, is needed to look for varices. An EGD involves inserting a small camera into the mouth down the food pipe (esophagus) to the stomach and the first part of the intestine (duodenum).

Some treatments can be performed through endoscopy to stop or prevent bleeding, such as banding (placing rubber band around varices) or sclerotherapy (injecting a substance into the varices). Some individuals with portal hypertension are prescribed blood pressure medications to reduce the pressure in the abdominal veins. In children with abdominal fluid collections (ascites), diuretics and a low-sodium diet are sometimes used to reduce fluid buildup. In children with portal hypertension and very enlarged spleens, the spleen can be at risk for rupture (tearing); they are usually advised not to participate in contact sports and to wear a spleen guard during physical activities.

Some children with severe complications from portal hypertension will need a surgical procedure called a portosystemic shunt. This procedure relieves pressure in the portal system by diverting blood from the portal system to other veins.

CHF and liver transplantation

About 5-10% of children (1 in 10-20) with ARPKD/CHF will need a liver transplant. The most common reasons for needing a liver transplant in CHF are severe portal hypertension (e.g. repeated bleeding from varices) or because of recurrent infections (e.g. cholangitis). Regular follow-up with a pediatric GI/liver specialist (hepatologist) will help to monitor for complications and help to decide if and when a liver transplant may be required.

Some children with ARPKD/CHF may need both a kidney and liver transplant. These can either be done one after the other, or at the same time, depending on the individual patient's needs. Good communication between the hepatology (liver) and nephrology (kidney) teams and the patient/family is imperative for proper planning and follow-up.

Medical follow-up of children with ARPKD/CHF

Given the variety of issues that can arise in children with ARPKD, many different specialists may need to be involved in their medical care. The child's **primary care physician (pediatrician)** can be extremely helpful to coordinate care between various medical providers and specialists.



Medical providers and specialists can include:

Neonatologists

Pediatricians specialized in newborn care

Pediatric nephrologists

Pediatricians specialized in the care of kidney diseases

Pediatric gastroenterologists

Pediatricians specialized in the care of digestive

Pediatric hepatologist

Pediatricians specialized in liver diseases

Pediatric surgeons, including transplant surgeons

Pediatric dietitians/nutritionists

Pediatric endocrinologists

Pediatricians specialized in growth and hormones

Developmental pediatricians

Specialists in learning and development

Occupational therapist, physical therapist, speech therapist, and feeding therapist

Geneticists and genetic counselors

Child life specialists

Providers who help children cope with illness, most likely through the hospital

Neurologists

Social Workers

Can help to obtain appropriate resources, navigate health insurance issues, etc.

Psychologists

Specialists in behavior and emotional well-being of children

Psychiatrists (if medication is ever needed for emotional issues surrounding living with a chronic illness or trauma)



Tests used to monitor health in children with ARPKD/CHF

Several tests are used to monitor the health of children with ARPKD/CHF. These can include blood tests, urine tests and imaging (radiology) studies. Commonly used tests are listed below, but there may be other tests that are needed, depending on an individual's needs.

Blood tests

Blood tests are used to monitor kidney function, electrolytes, liver function and blood counts.

Kidney function

Creatinine is the preferred measure of kidney function. Creatinine is a normal waste product that is made by the muscles, then is filtered by the kidneys and excreted into the urine. If kidney function is abnormal, the body cannot get rid of creatinine as efficiently, and the blood creatinine level gets higher. A higher creatinine level means worse kidney function.

Normal levels of creatinine depend on body size and muscle mass; a smaller child will normally have lower creatinine levels than a larger child, teenager, or adult. The creatinine level is used to calculate **glomerular filtration rate (GFR)**, the measure of kidney function. Another marker of kidney function is **blood urea nitrogen (BUN)**, which is a normal waste product of dietary protein. BUN also increases in children with abnormal kidney function. It is also affected by protein and water intake. Cystatin C is a newer marker of kidney function and is also used to help calculate GFR.

Electrolytes

Important electrolytes that are measured on blood tests include **sodium, potassium, bicarbonate, calcium** and **phosphorus**. Some children with ARPKD can have low sodium levels. Other electrolyte problems that can develop in children with kidney disease include high potassium, low bicarbonate, low calcium or high phosphorus levels. Sometimes, medications or dietary changes are needed to manage these electrolyte problems.

Liver function

Liver function tests are often normal or may be slightly abnormal in children with ARPKD/CHF. Other tests such as blood counts or ultrasounds are often used to monitor liver disease. Liver disease can sometimes affect the body's ability to absorb vitamins such as vitamin D and vitamin K. Blood tests are sometimes used to measure these vitamin levels so that supplements can be prescribed if needed.

Blood counts

A complete blood count (CBC) includes **white blood cells (WBC)**, the cells that fight infections), **hemoglobin** (a measure of **red blood cells**, the cells that carry oxygen throughout the body), and **platelets** (the cells that help blood to clot).

Children with abnormal kidney function can have **anemia** (low hemoglobin levels) because of low iron levels and/or because the kidneys may not be making enough erythropoietin (EPO).

Children who have portal hypertension due to ARPKD/CHF can have low platelet counts and sometimes low WBC and anemia because of an enlarged spleen that breaks down blood cells.

Urine tests

Urine tests are often done to look for protein in the urine (**proteinuria**). Proteinuria can be a marker of kidney damage. Sometimes, medications are prescribed to decrease proteinuria. Urine can also be tested for the presence of WBCs, which can indicate a urinary tract infection (UTI). If a UTI is suspected, a **urine culture** can be done to look for bacteria.

Imaging tests

Ultrasound

Ultrasound is the most common imaging test to look at the kidneys and liver in children with ARPKD. It is a safe and painless test that does not involve any radiation. Ultrasound can be used to measure the size of kidneys and liver, and to look for cysts or dilated bile ducts. Ultrasound is also helpful to monitor the size of the spleen and blood flow in the portal system in children with portal hypertension.

Magnetic resonance imaging (MRI)

A special type of **MRI**, called magnetic resonance cholangiopancreatography (MRCP), is sometimes used to get more detailed pictures of the liver and bile ducts in children with ARPKD. MRI of the kidneys is performed less commonly, but is sometimes performed in research studies.

An MRI does not involve radiation, and instead uses strong magnets to produce a picture. Because of this, MRIs cannot be performed on people with certain types of metal implants. Sometimes, an intravenous (IV) contrast agent called gadolinium is given during an MRI. However, gadolinium should not be used in people with very low kidney function.



Echocardiogram

An **echocardiogram** is an ultrasound of the heart. It is commonly done to monitor heart health in individuals with high blood pressure (hypertension).



ARPKD/CHF research

Many researchers around the world are trying to learn more about ARPKD/CHF. Scientists still do not have a clear understanding of why kidney cysts or liver fibrosis develop. They are also still studying what can be done to prevent these problems. Ongoing research in ARPKD includes **laboratory-based research** to learn how ARPKD/CHF affects cell functions and pathways, and also includes **registries, databases, and observational studies** that gather information about patients with ARPKD/CHF. All of these research studies are important to help us understand the biology of ARPKD/CHF so that treatments can be developed in the future.

There are currently no approved treatments that can prevent or slow the effects of ARPKD/CHF on the kidneys and liver. However, there are several drugs that have been studied in animals. A clinical trial for a new drug in children with ARPKD/CHF started in 2017, and there are likely to be clinical trials of other drugs in the future. Visit clinicaltrials.gov for updated information about ongoing research studies.

Family impact of ARPKD/CHF

Aside from its impact on the affected child, ARPKD/CHF can also have many impacts on a family.

The stress from appointments, missed work, and the stress of trying to help your child stay on track educationally and socially has its challenges. Many support services and help can be obtained from the hospital, public school system, and family and friends who are champions for the child/ren and family can be helpful. Many times, it is finding the right support for your family that will make the difference.



Screening of siblings for ARPKD/CHF

After a child is diagnosed with ARPKD/CHF, many families face the decision of whether to test other children to look for the disease. It is often helpful to screen siblings for ARPKD/CHF by performing a kidney/liver ultrasound, because some children can have ARPKD without having significant symptoms. This can allow the child to get appropriate medical follow-up as needed.

However, some families choose not to screen other siblings for ARPKD because they worry about the anxiety it will cause, or about how the diagnosis will impact insurance or other issues.

It is a very personal decision if families choose not to have siblings tested for ARPKD. However, it is still important for siblings to be seen by their primary doctor regularly and to have blood pressure checks and urine tests to look for complications of ARPKD.

Many families elect to make sure health and life insurance issues are settled before any potential diagnosis takes place.

Some families report that addressing a diagnosis early is better in the acceptance of treatment and management. In addition, some families feel that withholding the information from children can subconsciously send the message that a diagnosis is shameful.

Taking care of yourself as a parent or caregiver

When a child is diagnosed with any long-term chronic disease, there may be feelings of anger and grief. At some point, a parent or caregiver usually moves into advocacy mode. With a chronic disease that impacts everything from physical health to mental health and social and educational well-being and family dynamics and interactions, it's important to care for yourself.

Practicing self-care takes dedication, hard work and creativity. Here are some tips:

- Build your family's community and ask for help. Make asking for help a priority.
- Lower your standards, where possible. It's okay if your home isn't as clean as it once was or as clean as you would like it. Spend less time worrying about the house and more time figuring out how to practice self-care.
- It's OK to be selfish. Really. It is.
- If things get intense and you cannot leave your child/children, invite someone over when you are at home. Having another adult there could give you an opportunity to take a bath or nap or to do something you enjoy such as baking, reading, writing, working on a puzzle or crafting.
- When the house is quiet and the kids are asleep, take some time for yourself. Do something for yourself. Something that makes you happy. Something that relaxes you.
- If you are partnered up, talk about how you can work this out as a couple and as a family. You are in it together.
- Do not let anyone tell you what you "should" be doing to relax. If cleaning and organizing is what makes you happy, then do it.
- Be committed to something for yourself. Similar to the hundreds of goals our kids have to meet over their young lives, we should have



Julia Roberts, pictured at left (far right) with her family, has two children living with ARPKD.



Michele Karl, pictured with her family at right, has three sons, two living with ARPKD/CHF.



goals of self-care. We should treat time for ourselves as important as doctor appointments for their care. This could include getting a regular manicure/pedicure, joining a book club, taking a lesson or attending a class, or seeing a movie. Have a date or friend date night regularly.

- Ask your friends and family members for support. They can help you with the kid/s, errands, join you in an activity or be some kind of accountability partner. The people who love you want you to be happy and focus on you a bit.

Be kind to yourself. Give yourself a break. Get over the need to do everything perfectly. We encourage you to find time for yourself and not feel guilty about it. Self-care in special needs parenting is possible.



Educational impact of a disease/ chronic illness

A chronic illness can impact a child and his or her family in many ways with ongoing symptoms and treatments. Two of the most invasive impacts can be educational and social, yet are unavoidable because health care treatment often takes precedence. Symptoms can sometimes present themselves slowly, over time, and can potentially create an emergent situation.

It is important to plan and prepare for the possibility that a child may be unable to attend school or for when treatment may impact a child's ability to participate in daily activities. Children may need help navigating their social world as well because missing school and activities can impact social interactions with friends.

Because ARPKD/CHF are progressive conditions and have varying degrees of physical impact at different ages, the educational and social needs can vary and require monitoring and intervention.

What's the difference between a 504 Plan and an IDEA (IEP)?

Children with disabilities have been protected by the Individuals with Disabilities Education Act (IDEA) since the 1970s. They have been entitled to a free and appropriate public education, promising reasonable access to education.

Children have different rights under Section 504 (504 Plans) and IDEA (IEP, or Individual Education Plan). The difference is that for an IEP the disability impacts their ability to learn and the 504 plan is for children who may need accommodations but for which their disabilities don't necessarily impact them keeping on a grade level.

	IEP	504
What is it?	A plan for no-cost special education services for a child. Based on the IDEA act and federal special education law.	A no-cost plan for access to learning at school. Based on Section 504 of the Rehabilitation Act of 1973 to prevent discrimination against people with disabilities.
Is your child eligible?	Must have one or more of specific disabilities (learning disabilities, including attention issues, some mental and intellectual disabilities, and some resulting in “other health impairment”). Those disabilities impact your child’s ability to learn from regular or general education.	The child has a disability (or condition such as ARPKD) that interferes with an education, but might not qualify for an IEP yet still needs accommodations.
Steps to IEP or 504	Testing can be requested by parents/guardians. If grades are impacted by suspected disabilities, the school is typically supportive.	Evaluations are not provided/paid for by the school systems for those seeking 504 plans. However, with conditions, some accommodations may be made.
Who writes up the accommodations?	The psychologist who tests the child provides recommendations, then meets with parents/guardians, special education and general education teachers.	A 504 plan can be requested and written by anyone (parents, principal, educators). If the 504 modifications are for medical needs, a doctor’s report may be required.
Plan specifics	Documentation with measurable goals and updates annually. You can modify/add an accommodation by requesting a meeting and discussing need and implementation.	Plans for 504 plans are distributed to the child’s educational team and are almost always less complicated than IEPs.
Parents’ rights	We are provided parents’ rights in writing. There is a process for disputes. Services are free.	We are provided parents’ rights in writing. There is a process for disputes. Services are free.
Reviews	Annual reviews with re-evaluation every three years.	Varies by state, and also three years to re-evaluate, if the plan demands it and is needed.
Plan accommodation examples	Typically, support with extended test time, reading tests to children, smaller workgroups, co-teachers or aides, plans on how to deal with difficult behavior, and homebound services	<ul style="list-style-type: none"> • Access to water all day • Access to restrooms all day (with freedom to go) • Air-conditioned classrooms and transportation. • Homebound services during transplant, if during school year.

Tips on working with educators

Parent-teacher relationships are built out of mutual respect. Planning, tenacity, communication, and giving are required to work well together.

It's easy to feel protective of our children (especially when parents and caregivers fight all day in various ways for their health). That can lead to some adversarial stances early on in relationships with educators, which isn't necessarily helpful in building a collaborative relationship.

Keep these tips in mind:

Respect their knowledge

Teachers have the experience in teaching. Be sure to begin your relationship with them by acknowledging that.

Put concerns in writing

Be prepared for meetings by providing a list of issues to discuss. This can help minimize the number of meetings needed.

Try to remain flexible

In setting up meetings, offer several time options out of respect for the teacher's schedule.

Praise educators often

If a teacher or staff member is going out of his or her way to help, let them know it is noticed. Share that recognition with their superiors.

Volunteer

Volunteering can be an excellent way to integrate yourself into the school system. It may help you navigate the channels to get what your child needs. It helps to be around the school during pick up and drop off to have conversations that may not otherwise happen.

Social issues

ARPKD/CHF and their symptoms can have great impact on a child's day-to-day life. Because of that, a child with ARPKD/CHF has very different childhood experiences. These issues may impact their ability to relate to others.

This list represents possible social issues to be aware of for children with medical needs. However, every child's social development is unique and influenced by many varying factors. Some children may have none of these issues and some may have many.

Maintaining friendships can be difficult because of missed time from school or regular life due to illness and appointments. To assist your child in growing friendships, it may be necessary to inform other parents about what your child is going through.

Therapy can be helpful for everyone in your family. Your hospital or center may have a Child Life Specialist who can help your child process their medical experiences. Please refer to the Emotional impact section beginning on page 47 for more information on how to help your child and family cope.



The freedom to talk about frustrations around these issues goes a long way to a child's acceptance. Offering your child control of aspects of his or her ongoing care and life, when you can, could help. For example:

- Allow your child some control in when and how meds are delivered and administered (i.e., in the kitchen, before dinner/ after dinner, where they sit, counting before or not counting, with a particular flavored item to follow up a bad tasting liquid.)
- Offer your child the opportunity to select one appointment-free day. Agree to not schedule any appointment on those days. You will be surprised how health care professionals do not mind working around that. (The exception could be when the kidney or liver clinic at the hospital is open only on specific days.)
- Ask your child their preferences for lab work. For example, which arm to use, if possible, to draw blood. Offer them the opportunity to ask the phlebotomist questions such as how long they have been drawing labs.
- If they are uncomfortable in their presence, give the option to change health care professionals, when possible.
- If they are on a specific diet, provide them options for what items they want to have on hand at all times. If they have to consume a recommended amount of water, devise a system with their input (i.e., what water bottle with lines on it, and chart system with rewards.)
- Brainstorm with your child for options of treats after particularly hard times of all-day appointments.
- Let them express their feelings about what they do not like about a treatment, medication or procedure. Come up with options to help give them control of part or all of the process.



Medication/Administration

The amount of medication and the ability to administer it can impact if a child can sleep over and attend events and parties. The question remains who will be responsible for the medication and the timing. Medication can often be liquid, which would need refrigeration, and pills as they are able to swallow them properly. Medication administered at school may cause embarrassment and may need to be handled as privately as possible.

Medication/Symptoms

Discuss potential medication side effects with your doctor.

Appointments

Multiple appointments can be overwhelming. They may require numerous appointments from multiple specialties for regular care as well as when they are sick and receiving treatments and monitoring. These take time away from traditional childhood experiences because there are simply not enough hours in a day. Missing important milestones such as play dates and extracurricular activities can have an effect on how a child relates to the world around them for the simple lack of experiences. Some children may appear more immature than their peers because of the lack of interaction and experiences.

Obviously, missed school can have a major impact on their education. It is important to monitor their development in education to see if intervention is warranted. You may ask for an assessment from your public school at any time.

Dialysis

Depending on how dialysis is performed (hemodialysis or peritoneal dialysis on pages 20-21), your child may not be around school and friends enough to build strong bonds. They need the opportunity to have typical experiences that allow them to learn how to navigate friendships and social situations. Their education can suffer and the need for educational intervention may be required through an evaluation for an IEP or a 504 plan.

Diet

What a child can eat, and at what age special diets become medically necessary because of ARPKD/CHF can impact social situations. This may add to a child feeling “different” because of their diseases. Many things that are outwardly noticeable feel unfair to them.

Hospital stays

Less time at home or at school because of illnesses due to a chronic illness can cause a child to miss events and feel out of touch with the day-to-day functions of school, in friendships and in their family.

Fatigue

Being tired can be difficult. With failing kidneys and livers, children can feel tired. They may not be able to participate in regular activities. Children may sleep more and expend all of their energy for learning. This can lead to them needing extra sleep in the afternoons, and therefore reducing typical childhood experiences.

Emotional issues

Some children have behavior issues that can be isolating, creating barriers to friendships. Some emotional issues could be related to stunted emotional maturity and social skills because of the impact of a chronic illness and the medication treatments. See Social Issues on page 43.

When raising a child with ARPKD/CHF, it is easy to let the disease progression take over the focus of a caregiver/family. There is an overwhelming amount of information and physical management associated with the disease. Because of this, educational and social issues are often a secondary concern for a caregiver and family to address. Monitoring the potential educational and social issues that may arise could offer the opportunity for a caregiver/family to address the problems and change the outcome for the positive.

Emotional impact of ARPKD/CHF in children

The psychological and psychosocial impact of PKD upon adults has been fairly well documented and researched. There are websites, blogs and other tools to support and connect adults with PKD who are actively self-managing. There is considerably less information on tap for children diagnosed with PKD and their parents and caregivers, even though the impact of the disease is at least as consequential for them as it is for adults. An early life diagnosis of PKD is emotionally distressing and socially disruptive, as well as physically painful and exhausting.

Although many of the emotional and psychosocial issues faced by individuals diagnosed with PKD are the same regardless of age, children have a naturally limited capacity to articulate their subjective experiences and to elicit the support that they need. It goes without saying that children are inherently less powerful and less experienced than the adults around them. Thus, parents and caregivers must be prepared to recognize difficulties and to advocate for their children with wisdom and determination. Parental advocacy at its best should validate a child's experiences and prepare that child to advocate for herself or himself.

Emotional consequences of ARPKD/CHF may include symptoms of both anxiety and depression. In children, such symptoms often manifest physically and behaviorally long before the individual has the self-awareness and the vocabulary to articulate what they are experiencing.

Let's tackle ANXIETY first

The physical experience of anxiety may include the following:

- Feeling restless or on edge
- Feeling jumpy or exhibiting an exaggerated startle response
- Shortness of breath
- Feeling faint or dizzy
- Sweaty palms
- Racing heart
- Chest pain or pressure
- Muscle tension
- Feeling shaky
- Nausea
- Diarrhea
- “Butterflies” in the stomach
- Hot flashes
- Chills
- Numbness/tingling sensations
- A sense of unreality, feeling ungrounded
- Fatigue
- Sleep disturbances (difficulty falling or staying asleep, nightmares).



Thought patterns tend to run along the lines of “What if ...?,” “Oh, no ...!,” “People will laugh at me,” “I am going crazy” and “I’m having a heart attack and dying.” The content of thought will depend upon the individual, but the general idea is catastrophe.

Sequelae may include difficulty concentrating or staying on-task, difficulties with memory, and depressive symptoms such as disturbances of sleep and appetite (either too much or too little), lethargy and hopelessness.

The **behavioral manifestations** of anxiety may include the following:

- Avoidance or escape (refusing to go to school or other activities or needing to leave school/activities early due to feeling unwell)
- Unhealthy, risk-taking or self-destruction (overeating, bingeing and purging, use of alcohol or drugs, acts of self-harm*, acting out sexually)
- Withdrawal from daily activities or limiting the scope of such activities (quitting school, clubs or teams; or asking to be home-schooled because of pressures inherent in regular school)
- Over-attachment to a person, place or object associated with safety (going to great lengths to avoid separation from a parent, pet, treasured object, or home)

** Self-harm is basically the inflicting of pain upon one's own body, even if it doesn't really hurt. Examples include (but are not limited to) the plucking of hairs from one's eyelashes, head or other parts of the body (not as a part of grooming); picking of skin with fingernails or other sharp objects; re-opening wounds or scabs; burning the skin; cutting the skin; hitting or punching an object until the knuckles bleed; and beating the head against a hard object (generally a wall or floor). Often, the pain felt in the act of self-harm is minimal, though real damage is being done. Subjective reports suggest that the emotional pain or numbness preceding an incident of self-harm are experienced as worse than the physical pain. Such reports further suggest that it is the physical pain (or the sight of blood or wounding) that "grounds" the person and soothes the emotional discomfort.*

Depression

The physical experience of depression may include the following:

- Lethargy, easily fatigued.
- Increased aches and pains, particularly in joints, limbs and back.
- No (or low) appetite.
- Gastrointestinal distress.
- A sensation of heaviness, weakness and/or “like moving through molasses.”
- Chest pressure, tightness, feeling trapped, sometimes described as “claustrophobic.”
- Bad temper and short fuse.
- Hypersomnia (sleeping too much) with low energy/lethargy when awake (or too much sleep feeling like not enough sleep).
- Emptiness, sometimes described as “hunger of the soul.”
- A sensation of overwhelm and exhaustion. “Having my insides on fire while I slowly drown” or “like running up a hill of mud, every bit of progress you seem to make, you just slide back down.”



Thought patterns associated with depression tend to reflect overwhelm and isolation. Examples include — but are not limited to:

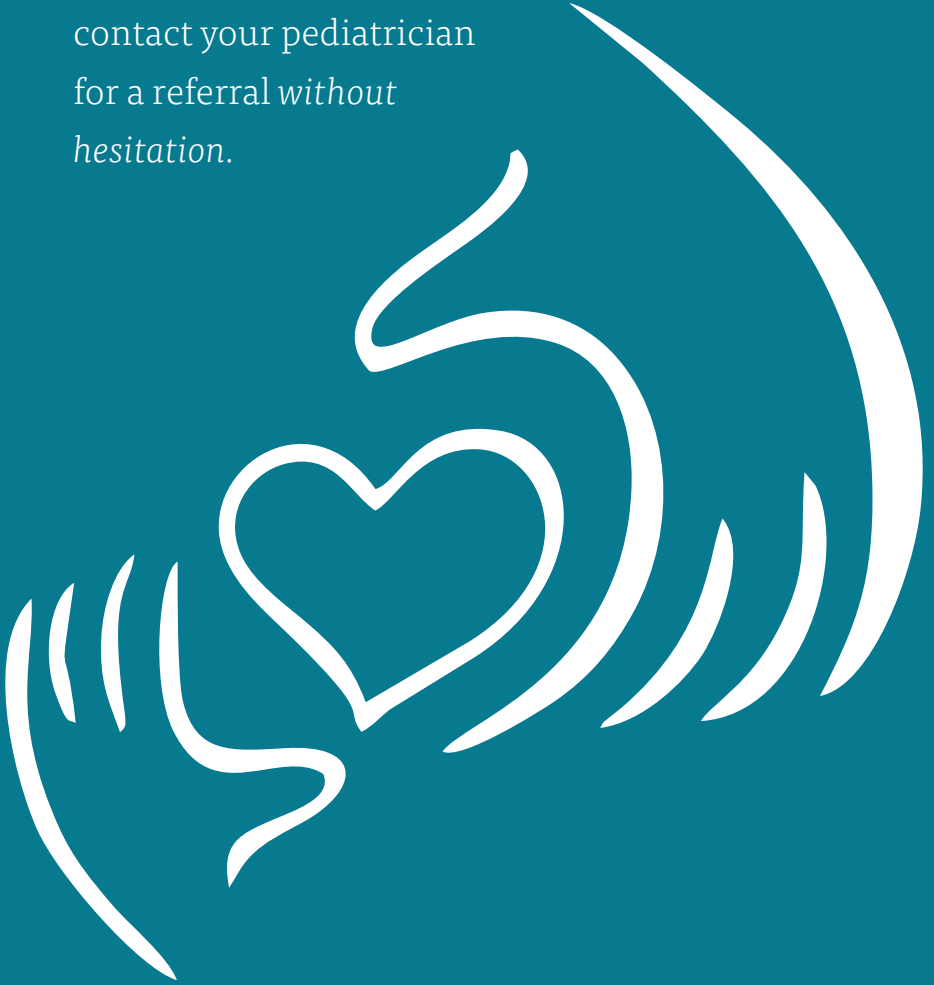
- “No one cares about me. The world is against me.”
- “There doesn’t seem to be a point to anything.”
- “I can’t care about anything, or I will get hurt.”
- “I cannot bear the effort of living.”
- “It’s all too much.”
- “I’ll be better off dead.”
- “Everyone will be better off without me to worry about.”

The behavioral manifestations of depression may include the following:

- They don't have the energy to deal with other people, so socially withdraw or quit what they can and limit the scope of anything that they are forced to do.
- They become apathetic or don't care about things that ought to matter to them.
- They seem to be going through the motions or are on autopilot.
- They withdraw from once-pleasurable activities, not due to maturation or a natural evolution of interests.
- They make statements reflecting low self-worth, hopelessness and helplessness.
- They show increased irritability and restlessness.
- They experience angry outbursts.
- They have trouble concentrating.
- They have difficulty managing the usual demands of school, work, home, family and friends.
- They exhibit poor hygiene. They do not care about cleanliness or appearance, refuse to bathe, brush teeth, and wear clean clothes.
- They harm themselves. See Self-harm on page 49.
- They threaten to kill themselves. Or they attempt to.

Sequelae may include serious injury, disability or death. In the long term, individuals with untreated depression may experience poor physical health, high risk for interpersonal conflict, increased risk of marital difficulties, financial impediments and compromised quality of life.

If you suspect there is suicidal ideation, threats and actions, call 911 or contact your pediatrician for a referral *without hesitation.*



What do you do if you notice a symptom from one of the lists?

First, if you suspect there is suicidal ideation, threats and actions, call 911 or contact your pediatrician for a referral *without hesitation*.

Second, don't panic. Like any of their other medical conditions and symptoms you manage, you have to find the right health care professional/s to help. It is good to start with play therapists for younger children, and therapists/counselors for older children. It can be beneficial to pursue family counseling if needed. Some of the ways you can support your child throughout their emotional journey are:

- Listen. Let your child express his or her fears and frustrations in whatever way they wish. Sometimes, children sense a parents' apprehension or fear and squelch their own out of concern for their parent.
- Use your child's interest to help them express their emotions. Examples are creating a story book of emotions and expressions using their favorite stuffed animals, setting up a special space if they like to color or draw, or through music or writing.
- If they express an interest in speaking with someone professionally, act quickly.
- Encourage friendships with kids that are compassionate (with compassionate parents) and who understand their need to come in and out of their friendships if needed, because of their health.

The impact of siblings

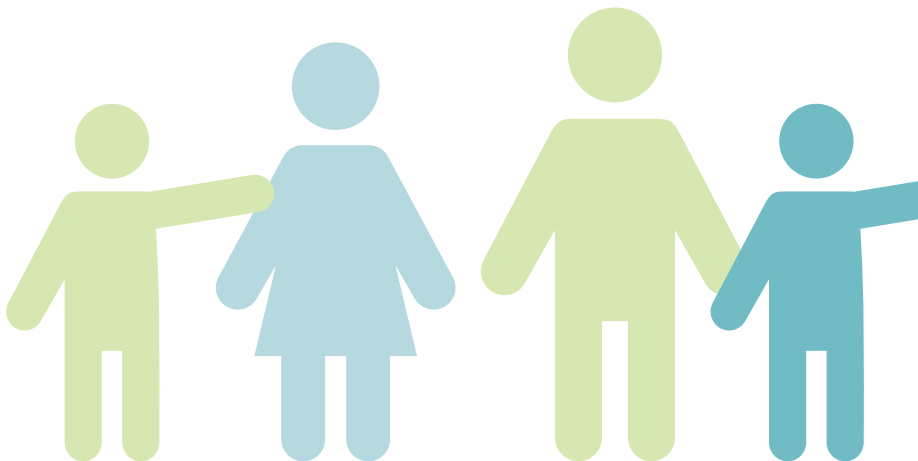
As a child or adolescent diagnosed with ARPKD references the family as his or her original and most important paradigm, the presence of siblings should be given some consideration. For a child or adolescent with ARPKD who has one or more siblings without ARPKD, there may be considerable and complicated emotions regarding the “unfairness” of things.

Sibling struggles and rivalries are normal and (usually) healthy manifestations of self-discovery. A child with a sibling is learning about the self and about the dynamics of relating to the world primarily through family. Relationships with peers are largely modeled on relationships with siblings. In the case of a child with ARPKD whose sibling is not forced to endure the pain and rigors associated with the disease, resentment can develop and the cumulative effect can be corrosive. Likewise, a sibling without the diagnosis could develop resentment over what could be perceived as an excess of attention being paid to the sibling with ARPKD.

The seriousness of the disease and the drastic measures that must be taken in the course of treatment mean that both the children with ARPKD and their siblings will be exposed to frightening ideas and experiences long before anyone is ready to process such things with them.

When parents and caretakers feel that sibling support is needed, one of the most effective means of providing this is through a play therapist. Play therapy can be effective for people of any age, but is the most effective treatment modality appropriate for children younger than 14. Many play therapists are also family therapists, and can incorporate other members of the family into treatment as needed.

When multiple children in a family have been diagnosed with ARPKD, trauma can be intensified for all family members. When siblings have the same disease, often one child will have more pronounced symptoms than the other. It is easy to see how a child (especially a younger child) whose symptoms are not yet so advanced could look to the case of the sibling and see his or her own eventual suffering. Of course, siblings who have been diagnosed can offer tremendous support to each other, even if only by being present as someone who understands well what the other must be going through. If a child wishes to be a support to a suffering sibling and this is something that the sibling is accepting of, then parents and caregivers can rest assured that this is not “putting too much” upon the comforter. Rather, such “in the trenches support” can benefit both siblings significantly. Adults can relate to the feeling of uplift and connection when “being there” for others in a time of crisis or need. Children and adolescents need such experiences to smooth the way into adulthood, regardless of medical diagnosis.



Unlike with knowing what is needed for kidney and liver failure (dialysis and/or transplant), mental health issues do not come with a roadmap, and there isn't a defined path of treatment.

What parents and caregivers can do

It seems inconceivable that a child would have to endure a disease of the kidney and liver and also suffer from mental health issues and maybe even trauma as a result of their medical treatment to keep them well and healthy. These lists of potential symptoms of anxiety and depression aren't intended as scare tactics. They are offered to prepare parents and caregivers so they may act swiftly in getting care for anyone in the family suffering from mental health issues, as they could manifest for patients with ARPKD, of course, but also caregivers and siblings.

Unlike with knowing what is needed for kidney and liver failure (dialysis and/or transplant), mental health issues do not come with a roadmap, and there isn't a defined path of treatment. Assessments from a team of professionals are warranted if you suspect any emotional issues. There are many routes to care and could include a psychiatrist assessment, counselors/psychologists, and play therapists for individual or family counseling. Additional considerations may be medication, journaling (for children too young to write, assist them in making storybooks) meditation, yoga, and exercise as a way to alleviate symptoms.

If medication is prescribed, the kidney and/or liver care teams should be consulted, as there can be medication interactions or adverse impact to the kidney or liver. Medication can be a hot topic issue. We strongly encourage that you work with a trusted health care provider to determine which treatments are best for the situation or family. It's also important to note that some medications do not work immediately, and some may need fine tuning and changing. It's a mixture of science and chemistry, so patience is needed while waiting for relief.



Resources

Now I Lay Me Down to Sleep

nowilaymedowntosleep.org

Now I Lay Me Down to Sleep is a 501(c)(3) non-profit that was co-founded by Cheryl Haggard, a woman who lost her baby shortly after making the excruciating decision to remove life support, and Sandy Puc', a photographer who came to the hospital to take pictures.

Support for Special Needs

supportforspecialneeds.com

Support for Special Needs is a site for essays, information and ideas by parents raising children with a variety of special needs. The co-founder's children have ARPKD and many posts are about her experiences.

Clinical trials information

- clinicaltrials.gov
- pkdcure.org/living-with-pkd/clinical-studies

Social Media

- **PKD in Children (PKD Foundation)**
facebook.com/groups/PKDinChildren
- **ARPKD**
facebook.com/groups/ARPKD
- **ARPKD Angels**
facebook.com/groups/arpkdangels

Glossary of terms

Access (hemodialysis)

The way to get blood from the patient to the hemodialysis machine.

Amniocentesis

A test used for prenatal diagnosis of genetic abnormalities. A small amount of amniotic fluid, which contains fetal cells, is sampled from the amniotic sac surrounding the fetus, and the fetal DNA is tested for genetic abnormalities.

Amniotic fluid

The protective fluid contained in the amniotic sac of a pregnant female; the fluid is partially supplied by fetal urine which is produced by the fetal kidneys; in ARPKD, poor prenatal renal function causes a reduction in this fluid.

Anemia

Commonly occurs in people with chronic kidney disease (CKD)—the permanent, partial loss of kidney function. Anemia might begin to develop in the early stages of CKD, when someone has 20 to 50 percent of normal kidney function. Anemia tends to worsen as CKD progresses.

Autosomal Recessive Polycystic Kidney Disease (ARPKD)

A multisystemic and progressive disorder characterized by cyst formation and “enlargement of the kidneys.” Occurs in approximately 1 in 20,000 live births.

Bicarbonate

A chemical in the blood that helps to balance the body’s acid levels. CKD can cause build-up of excess acid in the blood, making blood bicarbonate levels too low. Bicarbonate supplementation is sometimes needed in individuals with CKD.

Blood pressure

A measurement of the force of the blood as it flows through the body.

Blood Urea Nitrogen (BUN)

A measure of kidney function; urea nitrogen is the waste product of dietary protein, so if the urea nitrogen builds up in the blood, it is a sign of decreased kidney function.

Breathing problems due to underdeveloped lungs

Prematurity is the main cause of underdeveloped lungs. Babies with poor prenatal kidney function can also have underdeveloped lungs due to low amniotic fluid levels. Babies whose lungs are not fully developed at birth may have breathing problems.

Calcium

A mineral supplied in the food we eat and from calcium supplements. Vitamin D and parathyroid hormone (PTH) help regulate how much calcium is absorbed and how much calcium the kidneys eliminate. Healthy kidneys turn vitamin D into an active hormone (calcitriol), which helps increase calcium absorption from the intestines into the blood.

Caroli syndrome

An inherited condition characterized by abnormal widening (dilatation) of bile ducts within the liver, the presence of bands of fibrous tissue in the liver (congenital hepatic fibrosis) and high blood pressure in the portal veins (portal hypertension). Caroli syndrome can occur as an isolated form, or as part of a disorder such as ARPKD.

Carrier

An individual who carries one copy of a recessive gene like that for ARPKD; they do not have the disease but can pass the mutation on to their offspring.

Cholangitis

A serious infection of the liver's bile ducts. Individuals with abnormal bile ducts are at risk for cholangitis because the slow flow of bile can predispose to infection. Symptoms include fever, chills, and abdominal pain. In some cases, the skin may turn yellow.

Chorionic Villus Sampling (CVS)

A test used in prenatal diagnosis of genetic abnormalities in which a sample of chorionic villi is removed from the placenta for testing.

Chronic Kidney Disease (CKD)

Lasting damage to the kidneys that can get worse over time.

Collecting ducts

The last part of a long, twisting tube that collects urine from the nephrons (cellular structures in the kidney that filter blood and form urine) and moves it into the renal pelvis and ureters. Also called renal collecting tubule.

Congenital Hepatic Fibrosis (CHF)

A liver abnormality common in children with ARPKD; it may eventually lead to enlargement of the liver and spleen.

Creatinine

A waste product of muscle metabolism; the level of creatinine in the blood is a measure of kidney function. A high creatinine level can be a sign of abnormal kidney function. Creatinine levels are used to calculate glomerular filtration rate (GFR).

Cystatin C

A protein produced by cells in your body; the level of cystatin C in the blood is another measure of kidney function. A high cystatin C level can be a sign of abnormal kidney function. Cystatin C levels can also be used to calculate glomerular filtration rate (GFR).

Deceased donor

Most often individuals who die from accidents, heart attacks or strokes, and their next of kin consent to organ donation.

Dialysis

A treatment used to replace kidney function in someone who has kidney failure. The two forms of dialysis include hemodialysis and peritoneal dialysis. Hemodialysis uses a machine to circulate a person's blood through a filter outside the body. Peritoneal dialysis uses the lining of the abdomen to filter blood inside the body.

Dilated bile ducts

Abnormal widening of the bile ducts, which are small tubes that drain the liver of bile.

Echogenic

When kidneys appear brighter than normal on ultrasound.

Electrolytes

Chemicals in the blood such as sodium, potassium, phosphorus, calcium, and magnesium. The kidneys help to regulate the levels of electrolytes in the body.

End-Stage Renal Disease (ESRD)

When normal kidney function declines and needs to be replaced by dialysis or transplantation; also known as kidney failure; typically considered to occur when GFR is at 10 or less.

Enlarged, bright-appearing “echogenic” kidneys

Kidneys that appear larger and brighter than normal on ultrasound.

Endoscopy

An examination of the inside of the body using a lighted, flexible instrument called an endoscope. This procedure allows doctors to examine the interior of a hollow organ or cavity of the body.

Erythropoietin (EPO)

A hormone made in the kidney that tells the bone marrow to make red blood cells; individuals with abnormal kidney function may require a synthetic EPO supplement.

Fluid balance

The kidneys help maintain electrolyte concentrations by filtering electrolytes and water from blood, returning some to the blood, and excreting any excess into the urine. Thus, the kidneys help maintain a balance between daily consumption and excretion of electrolytes and water.

Gastric (G) tube

A gastrostomy tube (also called a G-tube) is a tube inserted through the abdomen that delivers nutrition directly to the stomach. It's one way doctors can make sure kids who have trouble eating get the fluid and calories they need to grow.

Genetic testing

DNA testing is available for PKD. There are two types of DNA tests: gene linkage testing and direct mutation analysis/ DNA sequencing.

Glomerular Filtration Rate (GFR)

The test used to check how well the kidneys are working; it estimates how much blood passes through the glomeruli each minute. Glomeruli are the tiny filters in the kidneys that filter waste from the blood.

Hematuria

The presence of blood in the urine.

Hemodialysis (HD)

A procedure that removes extra fluid, electrolytes and waste from blood using a dialysis machine.

Hemoglobin

A red protein present in red blood cells, which is responsible for transporting oxygen in the blood.

In Vitro Fertilization (IVF)

A medical procedure where an egg is fertilized by sperm outside the body. Used in conjunction with PGD.

Kidney transplant

A surgical procedure to place a healthy kidney from a live or deceased donor into a person whose kidneys no longer function properly.

Living donor

When a living person chooses to donate their kidney (or other organ) to someone who needs a transplant.

Magnetic Resonance Imaging (MRI)

A screening test that uses a powerful magnetic field, radio frequency pulses and a computer to produce detailed pictures of the inside of the body.

Mutation

An unintended change or “typo” in a person’s genetic code.

Nasogastric (NG) tube

A tube that is inserted through the nose, down the throat and esophagus, and into the stomach. It can be used to give drugs, liquids, and liquid food, or used to remove substances from the stomach.

Paired donation

A transplant option for candidates who have a living donor who is medically able, but cannot donate a kidney to their intended candidate because they are incompatible (i.e., poorly matched). It consists of two or more kidney donor/recipient pairs whose blood types are not compatible; the two recipients trade donors so that each recipient can receive a kidney with a compatible blood type.

PD catheter

A flexible plastic tube that allows dialysis fluid to enter the abdominal cavity, dwell inside for a while, and then drain back out again. PD catheter placement is considered a minor operation, and complications are rare.

Peritoneal Dialysis (PD)

A type of dialysis that removes extra fluid, electrolytes and waste using the lining of the abdominal cavity.

Phosphorus

An electrolyte whose level can become too high in individuals with kidney disease. High phosphorus levels can cause damage to your body. Extra phosphorus causes body changes that pull calcium out of your bones, making them weak.

Platelets

Tiny cell fragments that circulate through the bloodstream and assist in clotting when there is an injury to the body.

PKHD1

The gene that codes for ARPKD.

Poor kidney function

The gradual loss of kidney function. Your kidneys filter wastes and excess fluids from your blood, which are then excreted in your urine. When chronic kidney disease reaches an advanced stage, dangerous levels of fluid, electrolytes and wastes can build up in your body.

Portal hypertension

High pressure in the veins around the esophagus, stomach and intestines, usually caused by liver disease. High pressure in these veins causes them to become dilated (enlarged). The dilated veins, called varices, can rupture and cause potentially life-threatening gastrointestinal bleeding. Portal hypertension can also cause the spleen to become enlarged, which can cause low red blood cell, white blood cell and platelet counts.

Potassium

An electrolyte whose level can become too high in individuals with kidney disease. High potassium in the blood is called hyperkalemia, and can cause symptoms such as nausea, weakness, numbness and slow pulse.

Pre-Implantation Genetic Diagnosis (PGD)

A form of early genetic diagnosis that uses in vitro fertilization. Eggs harvested from a mother are fertilized in a laboratory with the father's sperm; then the fertilized embryos are tested for ARPKD. Embryos that are diagnosed as free of the disorder are then placed in the uterus with the intent to initiate a pregnancy.

Primary care physician (Pediatrician)

A specialist in Pediatrics, Family Medicine, or Internal Medicine who provides general medical care to a patient, and takes continuing responsibility for providing and coordinating the patient's comprehensive care.

Proteinuria

Protein in the urine.

Recessive

A genetic characteristic that is only expressed when it is inherited from both parents.

Red Blood Cells (RBCs)

The cells in the blood that are responsible for carrying oxygen throughout the body.

Sodium

An electrolyte in the body whose level is regulated by the kidneys. Excess sodium (salt) intake can cause fluid retention and high blood pressure.

Splenomegaly

Abnormal enlargement of the spleen.

Ultrasound

An imaging method that uses sound waves to take pictures of the inside of the body; the most common and least costly imaging method for PKD.

Urine culture

A test that can detect bacteria in your urine. This test can find and identify the germs that cause a urinary tract infection (UTI).

Varices

Abnormally dilated veins in the esophagus (tube that runs from the throat to the stomach) or the stomach.

Vitamin D

A vitamin that is important for bone health. It is present in certain foods (such as dairy products) and is made in the skin when exposed to sunlight. Vitamin D needs to be activated by the kidneys. Abnormal kidney function can cause low levels of activated vitamin D. This can lead to abnormal levels of calcium, phosphorus, and a parathyroid hormone (PTH), which can cause problems with bone health.

Waste products

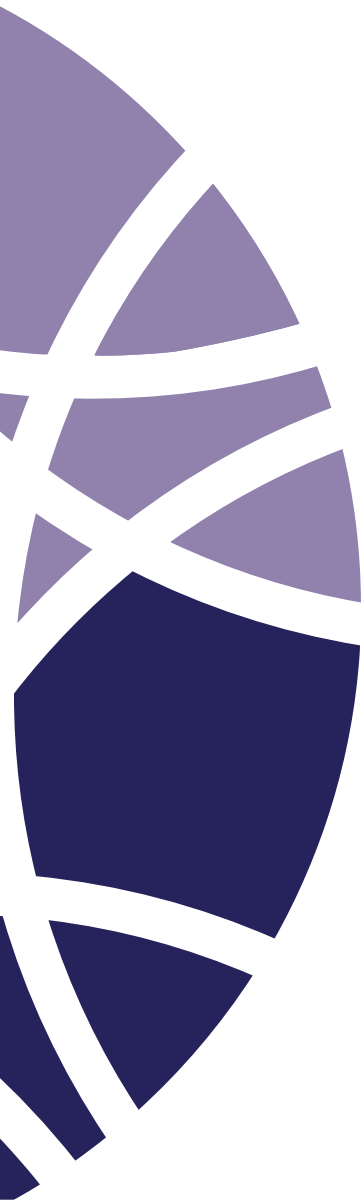
The kidneys remove waste products from metabolism, such as urea, uric acid, and creatinine, by producing and secreting urine.

White Blood Cells (WBC)

White blood cells in the blood help to fight infections. The presence of large numbers of WBC in the urine can be a sign of a urinary tract infection (UTI).



pkdcure.org/arpkd



PKD FOUNDATION
Polycystic Kidney Disease

pkdcure.org
1.800.PKD.CURE