



ARPKD PATIENT HANDBOOK

Your Guide to Understanding and Living with Autosomal Recessive Polycystic Kidney Disease and Congenital Hepatic Fibrosis



PKD FOUNDATION
Polycystic kidney disease

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How to Use this Guide

This guide is designed to help anyone who is impacted by **autosomal recessive polycystic kidney disease (ARPKD)** and **congenital hepatic fibrosis (CHF)**. This includes:

- Parents and daily caregivers
- Siblings
- Teens and adults living with ARPKD
- Extended family members (grandparents, uncles, aunts, cousins)
- Others who work with and support the child (teachers, coaches, babysitters, etc.)

While this guide is **not** intended for those who are supporting someone with **autosomal dominant polycystic kidney disease (ADPKD)**, some of the content is applicable as it relates to raising a child with PKD. You can find our ADPKD Handbook at **pkdcure.org/handbooks**.

This guide includes information that is helpful whether the person with PKD was recently diagnosed or has been living with ARPKD and CHF for many years. It's designed to help you better understand:

- ARPKD, CHF, and their related medical symptoms
- How ARPKD and CHF are diagnosed
- Best ways to manage ARPKD and CHF symptoms
- Tips on how to work well with your child's doctor (or your doctor if you're an adult) and care team to make sure you're getting the best care available
- Parenting challenges you might face and some ideas to try
- Social and emotional challenges you or your child might face and how to help
- School and learning challenges you might see and how to support your child

Tips for Getting the Most from This Guide:

1. Scan through the table of contents and identify the information that is most helpful for you right now.
2. Pay attention to **bold** words and phrases throughout this guide. If a word is in **teal and bold**, that means a definition is provided in the Glossary of Terms in Appendix G on page 57. If a word is **bold and black**, that means it is important information being highlighted in the section.
3. Use the blank pages at the back of this guide to jot down questions as you read. Bring these with you to your next doctor's visit so you can ask your care team.
4. It is okay to reread sections. Some of the content is complex and it will take time to fully understand it.



Understanding the Basics of ARPKD

What Is ARPKD?

Autosomal recessive polycystic kidney disease (ARPKD) is a rare genetic condition that mainly affects the kidneys and liver. It's usually diagnosed in infancy or early childhood, though some cases may be detected before birth. In rare cases, it can be diagnosed in teens and even in adults. Approximately 1 in 20,000 children, teens, and adults will be diagnosed with ARPKD.

How Does ARPKD Affect the Body?

Impact on kidneys: People with ARPKD develop fluid-filled sacs (cysts) in their kidneys. Often, these cysts are tiny, so individual cysts may not be seen on imaging (such as ultrasound). These multiple small cysts make the kidneys larger than normal and can make it harder for them to remove waste from blood. This can lead to high blood pressure and even **kidney failure** in severe cases.

Impact on liver: Everyone with ARPKD also has some degree of **congenital hepatic fibrosis (CHF)**, which affects the small **bile ducts** within the liver and can cause scarring. CHF can also affect blood flow inside the liver. (For more on CHF, see page 24 in this guide.)

Whole-body impact: Some babies with ARPKD can have breathing problems at birth because the lungs may not fully develop. Many individuals with ARPKD have high blood pressure, which can be treated with medication. Some children can experience issues with growth and nutrition, which can be managed with medications and supplements.

What Causes ARPKD?

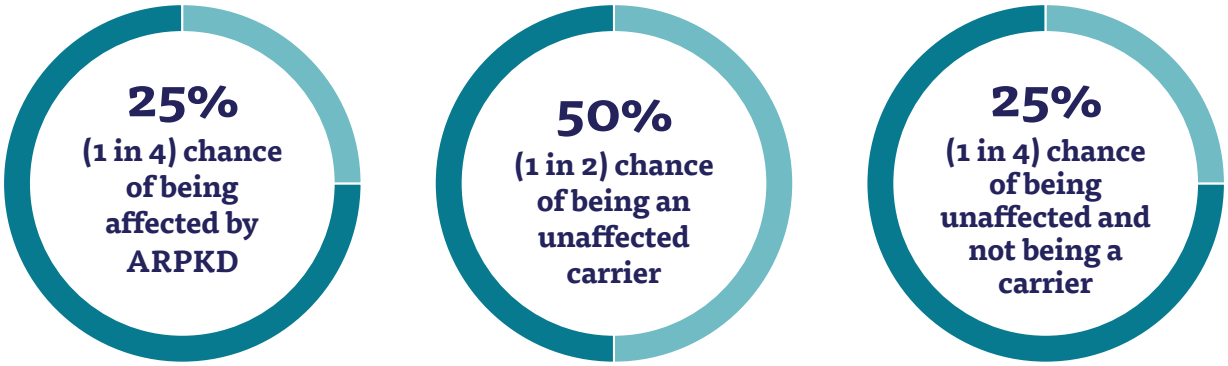
ARPKD is a genetic condition. It's most commonly caused by a **variant** (also called a **mutation**) of the *PKHD1* gene. This gene is important for proper development of the kidneys and liver.

ARPKD is inherited in an **autosomal recessive** manner. This means a child must receive a *PKHD1* gene variant from both parents to develop the disease. Both parents are carriers of the abnormal *PKHD1* gene but do not have the disease themselves. So, a person with one normal gene and one abnormal *PKHD1* gene will only be a carrier and will not have any symptoms of ARPKD.

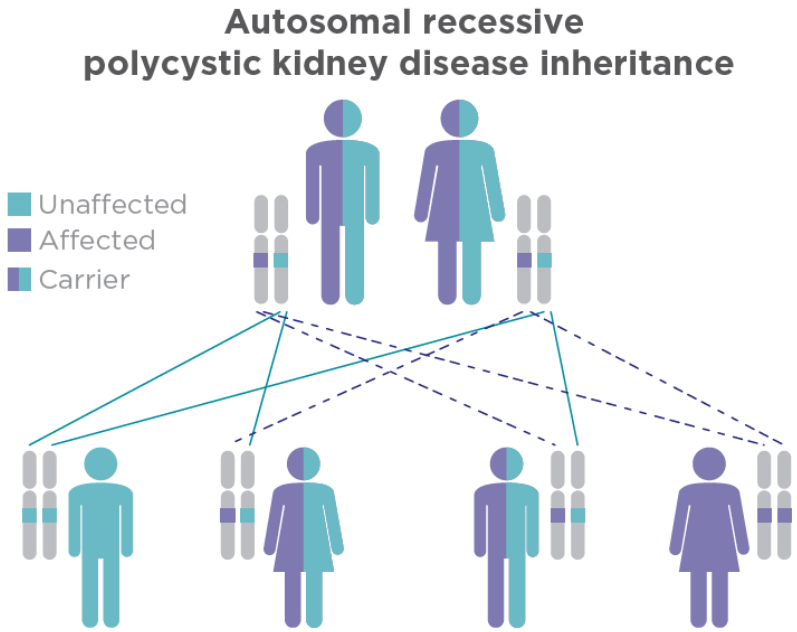
ARPKD can affect people of all races, ethnicities, and genders.

When both parents carry an abnormal *PKHD1* gene, there's a chance that a child will develop ARPKD, be only a carrier, or will be neither.

Here's how that works:



Most people don't know they're carriers of ARPKD until they have a child affected by ARPKD. There are many different types of *PKHD1* gene variants. The symptoms of ARPKD don't always correlate with the type of gene variants, and sometimes, siblings with the same gene variants can have very different ARPKD symptoms.



Does ARPKD Affect Everyone the Same Way?

No. Symptoms and severity can vary widely from person to person—even within the same family. Some people have more severe **kidney problems**, while others have more severe **liver problems**.

What Does an ARPKD Diagnosis Mean for Your Child?



The impact of ARPKD on each child varies greatly, even with children who have the same gene variants. Here are some basics to consider as you talk with your healthcare team. Keep in mind that each child and each ARPKD journey is unique.

- ARPKD can be diagnosed and start showing symptoms in infants or even before birth.
- Some children are diagnosed much later in childhood.
- Infants diagnosed before or soon after birth often have poor kidney function and underdeveloped lungs that require immediate treatment.
  - Nearly 8 in 10 babies who are diagnosed with ARPKD before or soon after birth will survive the newborn period.
  - If a child with ARPKD responds well to treatment during the first month of life, the outlook for survival is very good.
- Overall, **dialysis** or a **kidney transplant** is needed in about 1 in 4 children with ARPKD by 10 years of age, and in about half of children (1 in 2) by 20 years of age.
- The chances of needing dialysis or kidney transplant depend on when ARPKD was diagnosed (see "Understanding Kidney Function and Dialysis" on page 19).

REVIEW: Key Points – ARPKD Basics

- **ARPKD is a rare genetic disease** that affects the kidneys and liver.
- **Cysts in the kidneys** can cause high blood pressure and kidney failure.
- **Liver scarring from CHF** can affect bile ducts and blood flow within the liver.
- **Both parents must carry a *PKHD1* gene variant** for a child to have ARPKD.
- **Symptoms vary**—Many patients have high blood pressure, and some have more kidney problems, while others have more liver problems. Some patients have mild symptoms and others may have more severe symptoms.



Diagnosing ARPKD at Different Ages

Many individuals with ARPKD are diagnosed before birth (prenatally), right after birth, or in early childhood, but some are diagnosed in their teens or in adulthood. A diagnosis of ARPKD can often be confirmed with genetic testing.

How Is ARPKD Diagnosed Before Birth (Prenatally)?

Prenatal Ultrasound

ARPKD is often first suspected based on a routine prenatal ultrasound. ARPKD may be suspected if:

- A prenatal ultrasound shows that kidneys are larger than usual and appear brighter than normal (echogenic) because they have many tiny fluid-filled cysts.
- The amount of **amniotic fluid** in the womb is lower than it should be because the baby isn't creating enough urine due to the kidneys not working properly.
- 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 a prenatal ultrasound. Therefore, it isn't always possible to make a clear diagnosis of ARPKD based on an ultrasound unless other children in the family already have a confirmed diagnosis of ARPKD.

A normal prenatal ultrasound doesn't always mean an unborn baby does not have ARPKD. Sometimes kidney abnormalities don't develop until later in pregnancy or even after birth.

Prenatal Genetic Testing

Since an ultrasound doesn't 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 carriers of ARPKD.

A procedure, such as **chorionic villus sampling (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. An 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've had a previous child who was severely affected with ARPKD.

After Prenatal Diagnosis: What Are the Next Steps?

Prognosis (Severity and how the disease might progress)

It's often hard for doctors to know how severely a baby with ARPKD will be affected based solely on a prenatal ultrasound. Genetic testing doesn't always help predict a baby's outcome or how ARPKD will progress because the type of *PKHD1* variants don't always correlate with symptoms.

Here's what parents should know:

- Sometimes, there are such severe signs on a prenatal ultrasound (for example, severely underdeveloped lungs or lack of amniotic fluid) that the chances of a baby surviving after birth may be very small.
- Other times, a baby who doesn't show signs of severe abnormalities could develop severe, life-threatening complications after birth.
- And there are times when a baby, who appears to have severe abnormalities on a prenatal ultrasound, is able to survive after birth.

Finding Guidance and Support After Prenatal Diagnosis

It can be overwhelming to receive an ARPKD diagnosis before birth as you're faced with many difficult decisions. It can be helpful to seek guidance from doctors at specialized medical centers who have experience caring for families affected by ARPKD. These doctors can include:

- **Obstetrician (OB)**: A doctor who specializes in the care of women during pregnancy and childbirth.
- **Maternal Fetal Medicine (MFM) Specialist**: An OB who specializes in the care of high-risk pregnancies.
- **Neonatologist**: A doctor who specializes in the care of premature babies or newborns with complex health conditions.
- **Pediatrician**: A doctor who specializes in the care of infants, children, adolescents, and young adults.
- **Pediatric Nephrologist**: A doctor who specializes in the care of kidney diseases in children.

- **Pediatric Gastroenterologist (GI)**: A doctor who specializes in the care of digestion and the digestive system diseases in children.
- **Pediatric Hepatologist**: A pediatric gastroenterologist with additional expertise in the care of liver diseases in children.

Others who can help include:

- Social workers, psychologists, and child life specialists
- Palliative care providers
- Advocacy groups, family, and friends
- The PKD Foundation's PKD Parents Community can connect you with other families who have similar experiences.

In cases where survival after birth is thought to be very unlikely, families may need to make a hard choice about whether to:

1. Terminate the pregnancy.
2. Continue the pregnancy and be followed closely by an OB (MFM specialist) who has experience taking care of women with high-risk and medically complex pregnancies.

What is Pre-implantation Genetic Diagnosis (PGD)?

In families with a previous child severely affected by ARPKD, or in couples planning to start a family who have a family history of ARPKD, PGD can be a valuable way to prevent a child from being affected with ARPKD. To carry out PGD, the parents must do genetic testing to find out which *PKHD1* variants 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.

Consider Making Care Plans Before a Baby's Birth

It's important for parents to think about a care plan before a baby's birth. This includes choosing the delivery hospital. If a family is looking for maximum medical support, choosing a delivery hospital with an advanced neonatal intensive care unit (NICU) and specialists experienced in taking care of babies with severe diseases like ARPKD is important. The hospital team should include neonatologists, pediatric nephrologists, and pediatric surgeons.

Here are two things to consider:

1. Before choosing a hospital, ask about their experience with performing dialysis in infants.
2. If the survival of the baby seems very unlikely after birth, there are times when the medical team will recommend not providing lifesaving or life-extending care. This is rare but should be discussed with the medical team ahead of birth (see Appendix B for additional resources on page 44).

Always remember: finding the right care team for you and your unborn baby is a very personal choice. It may mean changing doctors or centers to find a care team that fully supports you and your family's wishes.

How Is ARPKD Diagnosed Immediately After Birth?

Sometimes ARPKD symptoms aren't evident until after birth. These symptoms can include:

- **Breathing problems due to underdeveloped lungs.** Some will require special support such as extra oxygen, continuous positive airway pressure (CPAP), or mechanical ventilation (breathing machine). Some may develop a pneumothorax (a collapsed lung).
- **Very large kidneys** that are pressing on the lungs, stomach, and intestines. This can cause breathing problems and make it hard for babies to eat.
- **High blood pressure (hypertension)** and low sodium levels (hyponatremia). These are discussed in more detail on page 14.



Sometimes, doctors will recommend that one or both kidneys be removed to make more room in the baby's abdomen to improve feeding and/or breathing. The potential benefits of removing the kidneys need to be balanced with the risks of surgery, the possibility that the infant will develop severe drops in blood pressure (especially if both kidneys are removed before 3 months of age), and the need for early dialysis. Dialysis is discussed on page 21.

Because of the complications described above, babies with ARPKD may have difficulty being fed by mouth. This may 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).

Every baby's care and situation is unique, so the above options need to be carefully considered and discussed with the care team.

How Is ARPKD Diagnosed in Children, Teens, and Adults?

Many individuals with ARPKD aren't diagnosed until later in infancy, childhood, or even adulthood. In general, individuals who are diagnosed with ARPKD after the newborn period tend to have less severe disease.

ARPKD may be suspected and diagnosed after a caregiver or healthcare provider notices symptoms like:

- An enlarged abdomen or kidneys that feel larger than normal (usually felt on the sides of the back)
- High blood pressure (hypertension)
- Urinary tract infections (UTIs)
- An enlarged liver or spleen that can be symptoms of liver disease
- Gastrointestinal bleeding (bleeding in the stomach or intestines)

Some individuals don't have these symptoms. They're diagnosed based on abnormal blood tests or abnormal imaging that was done for unrelated reasons or because they have a sibling with known ARPKD (see screening of siblings for ARPKD discussed on page 13).



What Diagnostic Tests are Used in Children, Teens, and Adults?

Several tests are used to help confirm a diagnosis of ARPKD. These include the following:

Laboratory Tests

ARPKD may be suspected based on abnormal blood or urine tests. However, the diagnosis must be confirmed using imaging and/or genetic testing, as described below. Lab tests that may suggest ARPKD include abnormal kidney function, electrolyte abnormalities (such as high potassium levels), or abnormal blood cell counts. See Appendix C for more about lab testing.

Imaging Tests

The most common imaging method to diagnose ARPKD is an ultrasound that provides an image of the kidneys. Kidneys in people with ARPKD generally appear enlarged and brighter than normal ("echogenic") and may have a speckled, "salt-and-pepper" appearance. This is because they contain many tiny fluid-filled cysts that are too small to be seen individually. A few cysts may be large enough to be seen individually and can appear as dark, fluid-filled bubbles, but these are usually <1 cm (1/2 inch) in diameter.

A scan of the liver may show uneven or irregular brightness, and the liver may be larger than normal. Some individuals may have visibly dilated (enlarged) bile ducts. If **portal hypertension** is present, the spleen may also be enlarged.

Genetic Testing to Confirm a Diagnosis of ARPKD

It's possible to test for ARPKD using **genetic testing** to look for variants in *PKHD1* and other genes. Testing will typically find a variant in about 8 in 10 (85%) affected patients. There are other childhood kidney diseases that can have similar symptoms to ARPKD. Most laboratories can test for multiple genetic kidney diseases at the same time. This can be helpful when the diagnosis isn't clear.

If a doctor thinks a child has ARPKD, testing can be done using a blood sample or sometimes using a saliva sample or cheek swab. 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's also possible to perform **pre-implantation genetic diagnosis (PGD)**, which is discussed on page 10.

Some affected individuals may have negative genetic testing or may only have one gene variant, rather than two (one from each parent). In these situations, the diagnosis of ARPKD is made based on imaging and clinical findings. However, there are other cystic kidney diseases that can look similar to ARPKD on imaging. Genetic testing often can detect those disorders as well.

Should You Test (Screen) Your Other Children for ARPKD?

After a child is diagnosed with ARPKD, it's important for families to think about the risk of the disease in their other children or in future pregnancies. Here's what you should know:

- Children born to the same couple will have a 25% (1 in 4) chance of being affected by ARPKD.
- Since children can have ARPKD without having clear symptoms, it's important to look for ARPKD in all other children including older ones. This allows them to get appropriate medical care if needed.
- Testing for ARPKD can be done by doing an ultrasound of the kidneys and liver. It can also be done with genetic testing, if the affected child's *PKHD1* gene variants are known.
- PGD can also be considered to minimize the chance of future pregnancies being affected by ARPKD. See more about pre-implantation testing on page 10.

Screening other children should be discussed with a qualified healthcare provider experienced with ARPKD.

REVIEW: Key Points – Diagnosing ARPKD

- **Diagnosis can happen before birth, at birth, or later in life**, depending on symptoms and family history.
- **Prenatal ultrasound** may show large, bright kidneys and low amniotic fluid but does not always confirm ARPKD.
- **Genetic testing during pregnancy** can help confirm the diagnosis, if parents are known carriers.
- **At birth**, babies may have breathing issues, large kidneys, or low sodium—diagnosis is made with imaging and labs.
- **In older children or adults**, diagnosis often follows signs like high blood pressure, UTIs, or enlarged organs.
- **Ultrasound and genetic testing** are the main tools to confirm ARPKD. Although, genetic testing may not detect all cases and some other genetic cystic kidney diseases can look like ARPKD on imaging.
- **Siblings may also be at risk** of ARPKD and should be screened.



Understanding and Managing Common Medical Challenges in ARPKD

All About High Blood Pressure (Hypertension)

Blood pressure (BP) is a measurement of the force of blood in the arteries as it flows through the body. The top number (systolic BP) measures the force of blood while the heart is contracting. The bottom number (diastolic BP) measures the force of blood while the heart is relaxing.

Understanding Normal BP in Children

BP in children is lower than in adults and naturally increases as a child grows. Normal BP values for children under 13 years old are based on a child's age, sex, and height, and are expressed as percentiles relative to a population of healthy children. For example, if a child's BP is at the **90th percentile, this means their BP is higher than 90 in 100 healthy children** of the same age, sex, and height.

The table below shows cut-offs for BPs that are considered elevated (90th percentile) for children of average height.

Note: *This table is for illustration purposes only. A child's actual cut-off values will vary based on their height. Please consult your child's doctor to understand their actual cut-off values.*

Simplified table of BP cutoffs for elevated BP (90th percentile) for children of average height *(based on the American Academy of Pediatrics 2017 Guidelines)*

Age, years	Boys	Girls
	90th percentile BP (You want a BP lower than this)	
1	100/53	100/56
2	102/56	103/60
3	103/59	104/62
4	105/62	106/65
5	106/65	107/67
6	107/68	108/69
7	109/70	109/70
8	110/71	110/72
9	110/73	111/73
10	112/74	112/73
11	114/75	114/74
12	117/75	118/75

Categories of BP

An individual's BP can be categorized as "normal BP," "elevated BP" (also known as pre-hypertension), or as "hypertension." This is based on repeated measurements over time.

In children younger than 13 years old, the categories are based on percentiles relative to children of the same age, sex, and height. In individuals aged 13 years or older, normal BP is based on standard cut-off values, as shown in the table below.

Definitions of BP categories in children and adults

Age	Younger than 13 years old	13 years old and older
Normal BP	Less than 90th percentile*	Less than 120/80
Elevated BP	90th- 95th percentile	120-129/less than 80
Hypertension	Greater than or at 95th percentile	Greater than or at 130/80

**90th percentile means the child's BP is higher than 90% of children of the same age, sex, and height.*

Treating Hypertension in ARPKD

Hypertension occurs in most (about 3 in 4) children with ARPKD and is caused by hormones that are released from damaged kidneys. BP can be difficult to control, particularly in babies with ARPKD. It's important to control BP properly to prevent complications, such as kidney damage, heart disease, and stroke.

It's common for individuals with ARPKD to need multiple medications to control BP. Examples of commonly used BP medications are shown in the table below.

Commonly prescribed BP medications used for children and adults

Class of BP medication	Examples of medication names (generic)
ACE inhibitors	captopril, enalapril, lisinopril
angiotensin receptor blockers (ARB)	losartan, valsartan
beta blockers	atenolol, carvedilol, metoprolol, propranolol
calcium channel blockers	amlodipine, isradipine, nifedipine
central alpha agonists	clonidine
diuretics	chlorothiazide, furosemide, hydrochlorothiazide, spironolactone

ACE inhibitors or ARBs are often preferred in individuals with ARPKD because they directly target the hormones released by the kidneys and have been shown to protect long-term kidney and heart health. However, these medications can sometimes cause high potassium levels so they may not be suitable for all individuals. Speak with your doctor about what medications may be right for you or your child.

Monitoring of Hypertension and Related Issues

To make sure an individual's BP is well controlled, doctors will sometimes ask families to check BP at home. It's important to use the correct technique, type of BP monitor, and cuff size to get accurate BP readings. Please consult with your doctor about the most appropriate BP monitor and technique for you or your child.

Doctors may also use a **24-hour ambulatory BP monitor (ABPM)** to diagnose hypertension or to see if hypertension is well controlled. An ABPM is generally provided by the doctor's office (nephrologist or cardiologist). It's worn continuously for 24 hours and automatically measures BP every 20-30 minutes while someone is awake and asleep. Because the child must be able to tolerate frequent blood pressure measurements, it may be challenging for younger children and/or those with developmental delays to undergo ABPM testing.



To make sure hypertension is not causing strain on the heart, doctors may also order an **echocardiogram** (ultrasound imaging of the heart). This test can look for thickening of the heart muscle in the main pumping chamber, the left ventricle (**left ventricular hypertrophy**), which can be a sign of poorly controlled blood pressure.

Low Sodium Levels in the Blood (Hyponatremia)

Low blood sodium levels often occur in babies with ARPKD. Sometimes, babies with ARPKD are treated with diuretics (medicines to help the body excrete water), feeding adjustments, and/or sodium (salt) supplements. Sodium levels generally normalize after infancy.

High Potassium Levels in the Blood (Hyperkalemia)

Individuals with poor kidney function can sometimes develop high potassium levels in the blood because the kidneys don't get rid of enough potassium. This can be treated using a specialized diet with decreased potassium. In infants, specialized low-potassium formulas can be used. If dietary changes don't control the potassium levels, medications may be used to help decrease potassium levels.

Urine Concentrating Abnormalities

Some children with ARPKD produce large amounts of urine because their kidneys are unable to concentrate urine well enough. Because of this, they may need more fluids than healthy children. This may also lead to bedwetting or the need for more frequent bathroom visits.

Urinary Tract Infections (UTI)

Individuals with ARPKD are at risk for UTIs, possibly due to slow urine flow in the cystic, dilated collecting ducts found in the kidneys. About 1 in 3 children with ARPKD will develop a UTI. These infections are more common in females. It's important to test for a UTI in anyone with ARPKD who develops painful urination, cloudy or foul-smelling urine, abdominal pain, and/or fever.

Nutrition, Growth, and Development Challenges

Children diagnosed with ARPKD in infancy, or early childhood, can have difficulties with feeding and physical growth. This can be caused by decreased food intake due to poor appetite and/or compression of the stomach and intestines by large kidneys, which can cause reflux or a sensation of early fullness. Some children may need a special feeding tube to help them take in enough nutrition and may require special formulas.

Decreased kidney function can also cause decreased appetite and growth problems in older children. Some children may need to take special dietary supplements to help them gain weight. Some children may also need growth hormone injections to help them grow.

A registered dietician or nutritionist can be an important member of the health care team for individuals with ARPKD. Below are some general nutrition principles to consider. However, **it's important that you get specific recommendations for your child from a dietitian or healthcare provider** since each child and situation is different.

Children with **normal kidney function or mild chronic kidney disease (CKD stage 1-3a)** generally benefit from:

- A **healthy, balanced diet** that focuses on low-fat sources of protein (such as fish, poultry, and nuts), fruits, vegetables, and whole grains, and avoids processed foods and junk food.
- A **low salt diet** to help control high blood pressure.
- Making sure the child **stays hydrated**. Many children with ARPKD can't concentrate their urine properly and will need more water than most children.

Some individuals with **moderate to severe chronic kidney disease (CKD stage 3b-5) benefit** from:

- **Restricting intake of electrolytes** such as potassium or phosphorus, if their kidneys can't get rid of them properly and their blood levels become high.
- **Decreasing their fluid intake**.

Note: *Not everyone needs the same dietary restrictions, so it's very important to discuss this with your medical team before making any dietary changes.*



Understanding Kidney Function and Dialysis

In those with ARPKD, kidney cysts make it difficult for the kidneys to work properly. Most individuals with ARPKD will eventually develop **chronic kidney disease (CKD)**. This means that the kidneys may not function properly and may not be able to get rid of the body's waste products.

In many people with ARPKD, kidney function worsens over time, eventually leading to **kidney failure**. If this happens, there are two options:

1. Dialysis
2. Kidney transplant

In ARPKD, the age that kidney failure happens varies widely from person to person. Overall, dialysis or a kidney transplant is needed in about 1 in 4 children with ARPKD by 10 years of age, and in about 50% of children (1 in 2) by 20 years of age. The chances of developing kidney failure depend on when ARPKD was diagnosed:

- For children diagnosed before birth, about 50% (1 in 2) will develop kidney failure by the age of 10 years.
- For those diagnosed after age one, about 10% (1 in 10) will develop kidney failure by the age of 10 years.

Symptoms of CKD can include:

- Tiredness (fatigue)
- Fluid retention
- Poor appetite
- Poor growth
- Problems with bone health
- Abnormal blood levels of electrolytes
- **Anemia** (low red blood cell counts)
- High blood pressure (hypertension)
- Learning/developmental problems

What do Kidneys Do?

Your kidneys are like the body's natural filters. Here's what they help with:

- Removing waste from your blood—things your body doesn't need—and sending it out through your urine.
- Controlling how much fluid is in your body, keeping just the right balance.
- Keeping important chemicals (electrolytes) like sodium and potassium at the right levels.
- Regulating your blood pressure by managing fluid and certain hormones.
- Making a hormone called **erythropoietin (EPO)**, which tells your body to make red blood cells.
- Activating **vitamin D**, which keeps your bones strong and healthy.

What Are the Stages of Chronic Kidney Disease (CKD)?

There are five stages of CKD, based on the level of kidney function. Kidney function is measured using the **estimated glomerular filtration rate (eGFR)**, which is calculated based on levels of creatinine, and sometimes cystatin C, in the blood.

Creatinine is a normal waste product that's made by the muscles, then filtered by the kidneys, and removed from the body in the urine. If kidney function is abnormal, the body can't get rid of creatinine and levels build up in the blood. A higher creatinine level usually means worse kidney function, but it's important to note that creatinine levels also naturally increase when a child grows due to increasing muscle mass.

Cystatin C is also a normal waste product made in most cells in the body and filtered by the kidneys. Similar to creatinine, a higher cystatin C level means worse kidney function. Cystatin C levels remain relatively stable as a child grows.

Doctors calculate an eGFR to estimate kidney function based on creatinine and/or cystatin C levels using equations that account for a child's age, sex, and height. The calculated eGFR provides the most reliable way to track and compare a child's kidney function over time.

CKD Stages

Stage	Description	eGFR (mL/min/1.73 m^2)	Kidney function (%)
1	Possible kidney damage (e.g., due to cysts) with **normal** kidney function	90 or above	90-100%
2	Kidney damage with **mild loss** of kidney function	60-89	60-89%
3a	**Mild to moderate loss** of kidney function	45-59	45-59%
3b	**Moderate to severe loss** of kidney function	30-44	30-44%
4	**Severe loss** of kidney function	15-29	15-29%
5	Kidney **failure**	Less than 15	Less than 15%

How Do You Manage and Treat CKD?

In the early stages, many of the symptoms and complications of CKD can be managed using the following approaches:

- Medications to manage electrolyte levels
- Blood pressure medications
- Vitamin D and iron supplements
- EPO injections to increase red blood cell count
- Growth hormone injections
- Diet changes

Once CKD progresses to stage 5 (kidney failure), treatments to replace kidney function, such as dialysis or a kidney transplant, are needed.

Dialysis Basics

Dialysis is a procedure that replaces some functions of the kidneys after they fail. It removes waste products and extra water and balances the level of different chemicals in the body, such as potassium and sodium (electrolytes).

Some babies need dialysis early in life if they're born with poor kidney function or if one or both kidneys require surgical removal. Other individuals may need dialysis later in life.

There are two main types of dialysis: **hemodialysis (HD)** and **peritoneal dialysis (PD)**. 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)

Hemodialysis, or HD for short, is a treatment that helps clean your blood when your kidneys aren't working well. A special machine works like an artificial kidney. It removes waste, extra water, and helps keep the right balance of chemicals in your body.

For HD, doctors need a way to move your blood from your body to the machine and back. This is called "**vascular access**."

- For young children and some older children, a soft tube (called a catheter) is usually placed in a large vein in the neck or upper chest.

For older children and adults, doctors can create a special blood vessel in the arm or leg by connecting an artery to a vein. This is called a **fistula** or **graft**.

HD is usually done in a hospital or clinic, but some people may be able to do it at home if the center offers that option.

Each HD session takes about **3-4 hours**, and most people need it **3-4 times a week**.



Peritoneal Dialysis (PD)

Peritoneal dialysis, or PD for short, is another way to clean your blood when your kidneys aren't working well. PD uses a special fluid called **dialysate** that goes into your belly to remove waste, extra water, and balance your body's chemicals.

For PD, a soft plastic tube (called a **PD catheter**) is placed into the belly in the space between the intestines and the wall of the abdomen during a small surgery. This tube stays in place and is used to fill your belly with dialysate fluid. While the fluid is in your belly, it pulls out waste and extra fluid from your body. After some time, the used fluid is drained through the same tube, and new, clean fluid is put in.

Each time the fluid goes in and out is called an **exchange** or **cycle**. Most people need multiple cycles every day. This is often done using a machine called a **cycler**, which works automatically at night while you sleep.

PD is usually done **at home**, either by the person receiving treatment or with help from a family member. It usually means fewer trips to the hospital or clinic compared to hemodialysis. It also makes it easier to attend school or work.

Possible Problems with Dialysis: What to Watch For

Both hemodialysis and peritoneal dialysis can sometimes cause problems. Doctors refer to these as complications. That's why it's important for children to receive dialysis care at a **specialized center** that has experience treating kids. These centers are better equipped to catch and treat any issues early.

Dialysis catheters (the tubes used in both HD and PD) need **very careful cleaning and care** to avoid infection. Even with careful care, there may still be a risk of infection. Sometimes, the catheters can get **blocked or move out of place**, which can stop them from working properly and may mean they need to be replaced. Some patients on PD may also experience abdominal pain, especially when they first start PD. This often improves with time and/or adjusting the dialysis prescription.

Dialysis also affects the body's fluid and chemical balance, which can cause side effects like:

- Feeling very tired
- Changes in blood pressure
- Gaining or losing weight
- Muscle cramps
- Feeling dizzy

That's why people on dialysis need **regular check-ups** to help manage these issues and stay as healthy as possible.

REVIEW: Key Points – Common Medical Challenges

- **High blood pressure (hypertension)** is common in individuals with ARPKD and can be hard to manage. It often requires multiple medications and careful monitoring.
- **Low sodium (hyponatremia)** and high potassium (hyperkalemia) levels may occur, especially in babies or children with poor kidney function, and may require supplements, diet changes, or medication.
- **Urine issues**, like large volumes or poor concentration, may cause frequent urination, bedwetting, and increased fluid needs.
- **Urinary tract infections (UTIs)** are more common in children with ARPKD, especially girls, and must be treated quickly to prevent complications.
- **Feeding and growth challenges** may occur due to poor appetite or pressure from large kidneys. Some children may need feeding tubes or growth hormone therapy.
- **Chronic kidney disease (CKD)** often develops over time, leading to symptoms like tiredness, poor appetite, bone issues, and anemia. Treatments include diet changes, medications, and, in later stages, dialysis or transplant.
- **Dialysis options**, including hemodialysis (HD) and peritoneal dialysis (PD), help replace kidney function in severe cases. Each has risks and requires close monitoring to avoid complications.



Kidney Transplant Basics

A **kidney transplant** is a surgery where a healthy kidney from a donor is placed into someone whose kidneys aren't working well. A kidney transplant generally results in better quality of life than being on dialysis. Because of this, transplant centers often try to plan a kidney transplant before someone needs dialysis, which is called a **preemptive transplant**. Children must be a certain size to receive a kidney transplant and usually need to weigh at least 10 kilograms (about 22 pounds). This means that younger children who develop kidney failure will require dialysis until they grow enough to be eligible for a kidney transplant.

The kidney can come from:

- A **living donor**, like a healthy parent, relative, or even a kind stranger.
- A **deceased donor**, through a national donor registry.

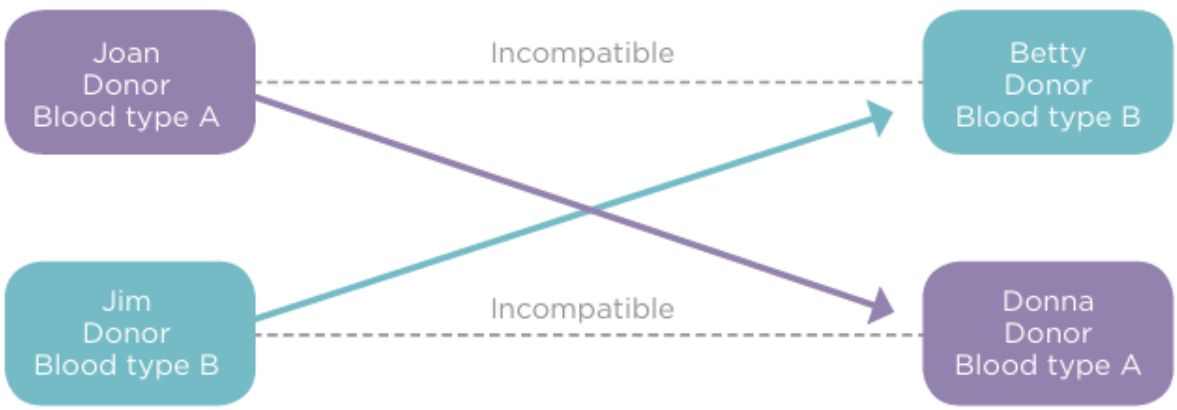
What is Paired Donation?

Sometimes a living donor wants to donate a kidney to someone they care about, but their blood types don't match. In that case, they can join a **paired donation** program.

In paired donation, two or more donor-recipient pairs "trade" donors so that each person gets a kidney matching their blood type. These exchanges can involve many families and take place across the country.

See the graphic below to understand how this works.

Paired Donation



What Happens Before a Transplant?

Kidney transplants are done at special hospitals with expert care teams. Before the transplant:

- Many **tests** are done to make sure the donor's kidney will be a good match.
- Living donors go through **health checks** to make sure it's safe for them to donate a kidney.

What to Expect After a Transplant

- After the transplant, you *must* take **medicine every day for as long as you have the kidney** to prevent the body from rejecting the new kidney.
- Some children with **ARPKD** may need to have one or both of their kidneys removed to make room for the new one.
- With good care, a transplanted kidney can work for a **very long time**. About **50% of children** who get a kidney transplant will still have a working kidney **20 years later**.

Will Another Transplant Be Needed?

Yes, most people who get a kidney transplant as a child will either:

- Need another transplant later in life.
- Need to go back on dialysis while waiting for a new kidney.

What If the Liver Is Also Affected?

- In some people with ARPKD, the **liver** is also seriously affected. These individuals may need both a **kidney and liver transplant**—either done one after the other, or both at once. Your doctors will decide what is best based on your specific needs.

Understanding Kidney Transplant Complications

A kidney transplant can be associated with various complications, and individuals who undergo a kidney transplant need close monitoring throughout their lifetimes.

After a kidney transplant, the body's immune system might see the new kidney as foreign and try to attack it. This is called **kidney transplant rejection**. To prevent this, doctors prescribe immunosuppressive medications that weaken the immune system. **It's crucial to take these medications exactly as directed**. Regular check-ups help monitor for signs of rejection so that any issues can be addressed promptly.

Infections can happen due to a weakened immune system. Because immunosuppressive drugs lower the body's defenses, individuals are more susceptible to infections. These can range from common colds to more serious illnesses. To reduce this risk:

- **Maintain good hygiene**: Wash hands regularly and avoid contact with sick individuals.
- **Stay up-to-date with vaccinations**: This helps protect against preventable diseases.
- **Be consistent with regular medical follow-ups**: Early detection and treatment of infections are vital.

Watch for liver-related complications. Individuals with ARPKD often have liver issues. After a kidney transplant, the risk of **cholangitis** (a bile duct infection) increases, especially if the person still has their own liver. Signs include:

- Fever (often with no other symptoms)
- Abdominal pain (sometimes)
- Jaundice (yellowing of the skin or eyes; this is rare)

If the symptoms of cholangitis appear, seek medical attention immediately. In some cases, doctors may prescribe preventive antibiotics to reduce the risk of cholangitis.

The Importance of Vaccinations Before and After Transplant

Before the Transplant

- **Complete all recommended vaccinations**: This includes vaccines for diseases like measles, mumps, rubella, and chickenpox.
- **Timing matters**: Live vaccines should be given at least **four weeks** before the transplant. Inactivated vaccines should be administered at least **two weeks** prior.
- **Accelerated schedules**: If the transplant is imminent, doctors might use an accelerated vaccination schedule to ensure maximum protection.



After the Transplant

- **Avoid live vaccines**: Post-transplant, live vaccines are generally not recommended due to the weakened immune system.
- **Inactivated vaccines are safe**: These include vaccines for influenza, hepatitis B, and pneumococcal disease.
- **Timing is key**: It's usually best to wait **three to six months** after the transplant before receiving inactivated vaccines, allowing the immune system to stabilize. However, the influenza vaccine can often be given as early as **one month** after transplant to ensure protection during flu season.
- **Regular monitoring**: Some vaccines may require booster shots. Regular blood tests can determine if additional doses are needed.

Protecting the Household

- **Vaccinate family members**: Ensuring that everyone in close contact with the child is up-to-date on their vaccinations helps create a protective environment.
- **Be cautious with live vaccines**: Household members should avoid live vaccines that can shed the virus, like the oral polio vaccine, to prevent potential transmission to the immunocompromised child.

REVIEW: Key Points – Kidney Transplant Recommendations

- **Kidney transplant recipients require lifelong monitoring** for complications such as rejection and infection.
- **Immunosuppressive medications are essential** to prevent rejection and must be taken exactly as prescribed.
- **Because immunity is lowered, infection risk increases**—good hygiene, staying away from sick contacts, and keeping up with medical visits are important.
- **Individuals with ARPKD may have an increased risk of cholangitis after transplant**; seek immediate care for fever, abdominal pain, or jaundice.
- **Complete recommended vaccines before transplant**; timing is important, especially for live vaccines.
- **After transplant, avoid live vaccines**; inactivated vaccines are safe but usually delayed 3–6 months (except flu vaccines, which may be given earlier).
- **Household members should stay fully vaccinated** and avoid live vaccines that can shed.



Understanding Congenital Hepatic Fibrosis (CHF)

What Is CHF?

Congenital hepatic fibrosis (CHF) is a liver condition that affects everyone with ARPKD to some degree. CHF happens because of the same genetic changes that cause ARPKD. It leads to scarring (also called fibrosis) in the liver.

Some individuals with ARPKD may never develop symptoms from CHF. Others, about 50%, may have mild to moderate symptoms during childhood or adolescence. A small number may develop more serious liver disease and need additional care or, in rare cases, a liver transplant.

What Symptoms Can CHF Cause?

CHF can affect different parts of the liver and nearby organs. Symptoms or problems may include:

- **Bile duct infections (cholangitis)**: The bile ducts in the liver may become enlarged and drain poorly. This makes it easier for infections to develop.
- **Portal hypertension**: This is when blood has trouble flowing through the liver from the intestines and spleen, leading to high pressure in nearby blood vessels.

It can cause:

- Enlarged veins (called varices) around the esophagus or stomach that can bleed.
- An enlarged spleen that may lower the number of blood cells, leading to:
  - Anemia (low red blood cells)
  - **Low platelets** (can cause easy bruising)
  - **Low white blood cells** (can increase infection risk)

How Is CHF Diagnosed?

CHF is part of ARPKD, but not all individuals will experience symptoms. Doctors use a combination of tools to check how the liver is functioning and for signs of portal hypertension.

1. Physical Exam

The doctor may look for:

- An enlarged liver or spleen
- A swollen belly
- Easy bruising
- Yellowing of the eyes or skin (jaundice)

2. Blood Tests

Blood work may show:

- Mild abnormalities in liver function tests (ALT, AST, bilirubin, or GGT levels)
- Problems with blood clotting
- Low white blood cells or platelets, if the spleen is enlarged

3. Imaging Tests

- Ultrasound is often used to look at the liver, spleen, bile ducts, and measure blood flow.
- **MRI** may be used to get a clearer picture of changes in the liver and bile ducts.
- Elastography can be used to measure liver scarring.

4. Liver Biopsy

Rarely, a small sample of liver tissue is needed to better understand the extent of fibrosis. This is usually done with a small needle and may require sedation.

How Is CHF Treated or Managed?

Regular Care and Monitoring

- Work with a liver specialist (hepatologist) if there are signs of CHF.
- Keep up with lab tests and imaging as recommended.
- Follow a healthy diet and exercise regularly to avoid fatty liver disease.

Nutrition and Vitamins

- Some individuals may need extra calories or vitamin supplements (especially A, D, E, and K) if their liver isn't producing enough bile to help digest food properly.
- A registered dietitian can help guide a nutrition plan.

Preventing Liver Damage

- Avoid alcohol.
- Make sure vaccinations for hepatitis A and B are up to date.



Medications

- Ursodiol may help bile flow better in individuals with enlarged bile ducts.
- Antibiotics may be needed to treat or prevent bile duct infections.
- Individuals who have had a kidney transplant, and are on immune-suppressing medications, are at higher risk of infection and should be watched closely for fevers or belly pain.

Managing Portal Hypertension

- **Endoscopy (EGD)**: This test involves putting a small camera into the food pipe (esophagus) and stomach to check for enlarged veins (varices) that could bleed. If needed, doctors can treat them during the procedure with banding or injections.
- Medications: Blood pressure medications may help reduce pressure in the abdominal veins.
- If an individual's spleen is very large, they may need to avoid contact sports or use a spleen guard to prevent injury.

Surgical Options

- In rare cases, individuals with serious portal hypertension may need a surgical shunt to lower pressure in the liver's blood vessels and reduce the risk of bleeding.

When to Get Urgent Help

Call your medical team or go to the emergency room right away, if you or your child experiences any of the following:

- Vomiting blood or passing dark, tarry stools (could be internal bleeding)
- Fever, particularly if accompanied by abdominal pain (could be cholangitis)
- Yellow eyes or skin (jaundice)

Liver Transplant in ARPKD/CHF

Most individuals with ARPKD and congenital hepatic fibrosis (CHF) will not need a liver transplant. But in some cases—about 1 in every 10 to 20 children (~10-20%)—a liver transplant may be needed.

The two main reasons for a liver transplant with CHF are:

- **Severe portal hypertension**, which can cause repeated episodes of serious bleeding from veins in the esophagus or stomach.
- **Repeated bile duct infections**, known as cholangitis, can be hard to manage over time.

To stay ahead of these issues, individuals should see a liver specialist (called a hepatologist) regularly. They'll keep track of the patient's liver health and help decide, if and when, a transplant might be needed.

In some cases, an individual may need both a kidney and a liver transplant. These transplants can be done either:

- **One after the other**
- **At the same time**, depending on what is safest and best for your child

It's extremely important that your liver and kidney doctors work closely together, and that they keep you involved every step of the way. This ensures the care plan is well coordinated and meets your child's specific needs.

Final Thoughts on CHF

While CHF is part of ARPKD, not all individuals will have symptoms or serious liver problems. With regular checkups and careful monitoring, most complications can be managed. You're not alone—your care team is there to guide you through every step.

Let your medical team know if you notice any new symptoms or have concerns. Early action can make a big difference.

REVIEW: Key Points – CHF Basics

- **CHF occurs in everyone with ARPKD** to some extent and results from the same genetic changes.
- **Symptoms vary widely.** Many individuals have no symptoms, but about 50% may experience mild to moderate issues in childhood or adolescence. A few may develop serious liver disease.
- **It's important to watch for complications.** CHF can cause bile duct infections (cholangitis), portal hypertension, enlarged spleen, anemia, easy bruising, and low immunity.
- **Diagnosis involves several tools** including physical exams, blood tests, imaging (like ultrasound or MRI), and, occasionally, liver biopsy to assess liver health and detect complications.
- **Ongoing monitoring is key for managing CHF.** It should include regular monitoring by a liver specialist, bloodwork, imaging, and a healthy diet.
- **Treatments are available to help manage chronic symptoms.** Medications, nutrition support, and procedures (like endoscopy or shunts) can help.
- **Know when to act fast.** Urgent care is needed for symptoms like vomiting blood, dark stools, fever with belly pain, or yellowing skin/eyes (jaundice), as these may signal serious complications.



Building a Strong Medical Care Team

Because ARPKD can affect many parts of the body, individuals may need care from several different medical specialists. A primary care doctor—such as a pediatrician, family doctor, or internal medicine doctor—can help coordinate care and keep all the different doctors on the same page.

Who Might Be on the Care Team?

In addition to a primary care doctor, specialists who may be involved at different times include:

Physicians:

- **Developmental pediatrician**: A doctor who helps with learning, behavior, and development.
- **Endocrinologist**: A doctor who helps with hormone problems and growth concerns.
- **Gastroenterologist**: A doctor who treats problems with digestion and the digestive system.
- **Geneticist**: A doctor who helps people understand how conditions like ARPKD are passed down in families and what that means for their health or future children.
- **Hepatologist**: A gastroenterologist with expertise in treating liver diseases. This doctor may be involved if the person with ARPKD has liver issues related to ARPKD.
- **Neonatologist**: A doctor who cares for newborns, especially those with medical issues.
- **Nephrologist**: A kidney doctor who helps manage ARPKD. Some nephrologists work with children (pediatric nephrologists), and some work with adults.
- **Neurologist**: A doctor who specializes in the brain and nervous system.
- **OB/GYN**: A doctor who provides gynecological and contraceptive care to women with ARPKD and supports the monitoring of healthy pregnancies in those expecting a child with ARPKD.
- **Psychiatrist**: A doctor who can help with emotional and/or behavioral challenges and prescribe medication if needed.

- **Psychologist**: Supports mental and emotional health, including behavior and coping.
- **Surgeons or transplant surgeons**: Doctors who perform surgeries like kidney or liver transplants, or who place feeding tubes and dialysis catheters.
- **Palliative care provider**: A doctor or nurse practitioner who provides symptom relief, care, and support to people living with serious or chronic illnesses.

Healthcare Team Members:

- **Child Life Specialist**: Someone in the hospital who helps children cope with being sick or undergoing procedures.
- **Dietitian or Nutritionist**: Helps with diet and nutrition plans to support growth and overall health.
- **Genetic Counselor**: Professional who explains how ARPKD is inherited and what it means for your family.
- **Home Healthcare Team**: A healthcare team who monitors individuals and performs complex medical treatments in their home.
- **Social Worker**: Helps with resources, navigating insurance, school needs, and family support.
- **Therapist**: This might include physical therapists (movement), occupational therapists (daily skills), speech therapists, or feeding therapists.

Working Together as a Team

Clear communication between you and your child's medical team is essential. For example, if one doctor is ordering blood work, they should check with the other doctors to see if any other tests can be added at the same time. This will minimize the number of required needle sticks. Some hospitals also offer appointments with multiple specialists on the same day to reduce the number of trips you need to make.

You Deserve to Feel Heard and Supported

You know yourself and/or your child best, and your voice matters. If a doctor doesn't listen to your concerns, or if communication isn't working well for you, it's okay to get a second opinion or switch to someone who makes you feel more comfortable and respected.

Not all doctors are familiar with ARPKD, especially in smaller or rural communities. That doesn't mean they can't provide great care, and it's important that they stay up-to-date on the latest research and treatment options. The PKD Foundation works closely with experts across the country to share information and support patients with ARPKD.

You can learn more or connect with the PKD Foundation at **pkdcure.org/arpkd-care**, or contact us at **pkdcure@pkdcure.org**.

The Role of an ARPKD Researcher on Your Medical Team

Some doctors who care for children, teens, and adults with ARPKD also do research to help find better treatments. These doctors are not only experts in treating patients but also in studying ARPKD to improve future care.

Many families choose to get a **second opinion** from one of these ARPKD researchers, especially soon after diagnosis. The **PKD Foundation** can help connect you with one of these experts through the **PKD HOPE Line** at **844-PKD-HOPE (844-753-4673)** or by emailing **education@pkdcure.org**.

Insurance companies sometimes cover the cost of a second opinion visit. However, it may not always be realistic for a family to use an ARPKD researcher as their regular kidney doctor (nephrologist).

The reasons for this could include:

- High costs if the doctor is not in your insurance network
- Long travel distances to see the specialist

Most families work with a **local nephrologist** close to home for regular check-ups and everyday care. They may visit an ARPKD researcher occasionally for help with more complicated issues, to learn about the latest treatment options, or to find out about research studies.

Even if you don't see an ARPKD researcher, you can still **learn about research studies that are looking for participants by going to this PKD Foundation's website at pkdcure.org/research/clinical-trials**.

Some research studies may involve traveling to the researcher's hospital or clinic. Often, the study will pay for travel costs. Other studies might only involve sharing your medical information and can be done from home.

Everyone impacted by ARPKD—children, teens, and adults—is encouraged to participate in research if they can. Research is an important way to help discover better care and new treatments for ARPKD in the future.





ARPKD/CHF Research and Hope for the Future

Researchers worldwide are working hard to better understand ARPKD and CHF. While we've made progress, scientists are still uncovering why kidney cysts and liver fibrosis develop in ARPKD and what can be done to prevent or slow these complications.

Current Research Efforts

Ongoing ARPKD research includes laboratory studies to explore how the disease affects cells and biological pathways. Additionally, large patient registries and observational studies are helping doctors and scientists track the progression of ARPKD and CHF over time. These studies are essential for advancing our understanding of the disease and identifying potential treatment options.

Hope for Future Treatments

Currently, there are no FDA-approved treatments to prevent or slow the progression of ARPKD or CHF. However, researchers are testing several promising drugs in laboratory models, and some are being evaluated in clinical trials for children with ARPKD. As research continues, we expect more clinical trials to emerge, offering hope for better treatment options in the future.

Finding Clinical Trials

If you're interested in learning about ongoing clinical trials (research studies) for ARPKD, you can visit:

- **ClinicalTrials.gov**: A database of publicly and privately funded clinical studies worldwide.
- **PKD Foundation Clinical Studies**: A resource dedicated to finding and participating in ARPKD research studies.

SIGN UP to learn when ARPKD clinical trials are seeking patients.

By staying informed and participating in research, when possible, patients and families can play an essential role in the search for better treatments.



Getting Ready to Move from Pediatric to Adult Care

Most young people with ARPKD will switch from pediatric doctors to adult doctors sometime between the ages of 18 and 21. But getting ready for this big step should start much earlier—often years before the actual change.

The goal is to help your child grow into a confident young adult who can take charge of their **own** health and keep getting the care they need.

Start Early: Ages 6–11

Even young children can start learning about their health. At this age, focus on:

- Explaining, in simple terms, that they have ARPKD and what that means.
- Talking about why they take medicine and go to the doctor.
- Helping them notice how they feel and when something might be wrong.
- Making healthy choices together, like eating well and staying active.

Building Awareness: Ages 12–14

This is a good time to start talking about the transition to adult care. You can:

- Ask your child's care team when they usually move patients to adult doctors.
- Find out what tools or support are available to help teens prepare.
- Start talking about the idea that, in the future, your child will be responsible for their own health.

Building Skills: Ages 14–18

Over these years, your teen can begin to take more responsibility. They should gradually learn how to:

- Explain their medical history and diagnosis.
- Manage their medications (like filling a pillbox or asking for refills).
- Make their own appointments.
- Talk directly with their care team. Start by having some time alone with doctors.
- Understand diet and lifestyle recommendations.
- Know what symptoms to watch for and what to do if they occur.
- Keep track of their medical records.

How you can help:

- Let them try doing things with your support first, then step back over time.
- Encourage decision-making. Ask what they think before you jump in with answers.
- Practice these skills together in everyday life.

Making the Switch: Ages 18–21

This is the time to complete the move to adult care. Here's what to expect:

- Choose a new primary care doctor (adult provider) to replace the pediatrician.
- Set up appointments with adult specialists, like a nephrologist and a hepatologist.
  - If you can't find an adult nephrologist who knows ARPKD, look for one with experience in autosomal dominant polycystic kidney disease (ADPKD).
  - The **PKD Foundation Centers of Excellence (pkdcure.org/findcare)** can be a helpful resource.
- Ask the pediatric team to send medical records and speak with the new doctors to ensure a smooth handoff.
- Check with your insurance company about coverage for the new doctors.

Important legal step: Once your child turns 18, you'll no longer automatically get access to their medical information. If they want you to stay involved, they'll need to sign a release form (also called a consent form).

You may also want to discuss setting up a **durable healthcare power of attorney**, which is a legal document that gives you the power to make medical decisions if they become unable to make decisions for themselves.

Final Tip

Consider scheduling a **final visit with the pediatric team** after the move is complete. It's a chance to wrap things up, ask last questions, and say goodbye.

Transitioning to adult care is a journey, not a one-time event. With time, practice, and support, your child can build the skills they need to thrive in adult care.

Other things to consider:

- **Where will your child be living at the time of the transition?** Will they be away at college for a period of time or at some other training or job site?
- **What are their ongoing medical needs?** You may need to decide whether dialysis or transplant should be done in the pediatric or adult system and consider the availability of specific expertise or services in the pediatric or adult system.
- **What is your child's level of comfort with an adult care model?**

Checklist: By the Time They Move to Adult Care

Your child should be able to:

- Describe their condition and treatment.
- Take medications on time.
- Track symptoms and know when to ask for help.
- Talk to doctors on their own.
- Manage appointments and health insurance.
- Decide who they want involved in their care.
- Bring their own medical info to appointments.
- Follow up after appointments and understand next steps.



Tips for Parenting Children with ARPKD or CHF

Parenting a child with ARPKD or CHF comes with unique challenges. Your child's experiences may look different from their peers. They may miss school, social events, or other activities due to medical needs and appointments. This can affect social and emotional development compared with peers of the same age.

In addition, the medical impact of ARPKD and CHF on the body can impact brain development, brain functioning, and learning. It's important for children with ARPKD to be evaluated for learning problems and to receive appropriate support in school, if needed.

Helping children feel in control of their life—when possible—can make a big difference. The practical advice below comes from parents and experts who understand what you're going through. Take what works for your family and adjust as needed.

Helping Your Child Build Friendships

Many kids with ARPKD or CHF struggle to maintain friendships because they miss school or social activities. You can help by:

- Encouraging playdates and virtual hangouts when in-person time isn't possible.
- Letting your child's friends, and their parents, know about your child's condition in a way that feels comfortable.
- Finding social opportunities that don't depend on physical energy, like art, music, or gaming.

Supporting Emotional Well-being

Living with a chronic illness is emotionally challenging for children and families. Counseling can help everyone navigate the ups and downs.

> **"I was worried my son would feel isolated, but we started inviting his friends over for short visits. It helped so much."**
>
> *– Parent of a child with ARPKD*

- Many hospitals have **child life specialists** who support kids emotionally, specifically during medical procedures. Ask for a child life specialist to be available when scheduling appointments.
- **School counselors** and **school nurses** can also provide support and guidance while at school and help provide resources/connections within their community.
- Talking about feelings—frustration, sadness, or fear—helps your child develop healthy coping skills.
- Let your child know that it's okay to be upset. Listen without trying to "fix" everything.
- Consider asking your primary care provider for referrals to individual or family counseling professionals.
- See Appendix F (on page 54) for more on how to support your child's emotional needs.

Giving Your Child a Sense of Control

Even small choices can help your child feel more in charge of their care.

- **Medications**: Let your child decide when and where they take them (e.g., before or after dinner, with a favorite drink).
- **Appointments**: If possible, let them pick one day a week that is appointment-free.
- **Blood draws**: Ask if they have a preferred arm or want to ask the lab tech a question first. Speak with your doctor to see if numbing cream is available before the blood draw.
- **Doctors**: Give your child a say in what healthcare providers they see. If your child is unhappy with a provider and switching providers isn't an option, help your child voice concerns about their care.
- **Diet and hydration**: If they're on a special diet, let them help choose snacks. If they need to drink more water, create a tracking system with rewards.
- **After long medical days**: Give them a choice of a small treat or activity afterward.

> **"Some days my daughter just needs to vent. I remind myself that listening is the best thing I can do."**
>
> *– Parent of a child with CHF*

- **Promote physical "can do's" when discussing physical limitations**: Some medical equipment or symptoms may limit certain physical activities, but it's just as important to encourage your child to safely participate in activities they enjoy. A physical or occupational therapist can help provide support and ways to help make this happen.

Medication and School Challenges

Medications can affect daily life, including school and social events. Some things to consider:

- Who will give medication at school, events, or sleepovers?
- Does the medication need to be refrigerated?
- If taking medicine at school is embarrassing, can it be done discreetly?

"We worked with the school to set up a 504 Plan so my son wouldn't fall behind. It was a game-changer."

– Parent of a child with CHF

Managing Appointments and Education

Frequent medical visits can be overwhelming and may cause your child to miss school and social events.

- Track school absences to ensure they're keeping up academically.
- Request an **educational assessment**, if needed. Public schools offer **504 Plans** and **IEPs** to help kids with medical conditions.
- If possible, schedule appointments at less disruptive times (e.g., afternoons or Fridays).
- Try scheduling appointments for labs and imaging tests in one trip.
- Check with the clinic to see if multiple doctors can be seen in a single visit.
- Check with the clinic to see if appointments can occur while on dialysis.
- Check with the clinic to see if appointments can be virtual.

Dialysis and Social Life

If your child is on dialysis, their school and social time may be limited.

- Look for ways to keep them connected with friends through video chats or short visits.
- If they miss a lot of classes, talk to their school about flexible learning options. Can they join class lessons virtually or can some lessons be recorded for them to view later?

Food and Feeling Different

Special diets can make kids feel "different" at meals and events.

- Pack familiar foods for outings, so they always have something they enjoy.
- Talk to your child about their diet in a way that focuses on what they can eat, not just what they can't.

Fatigue and Rest Needs

ARPKD and CHF can cause fatigue. Your child may need extra sleep or breaks during the day.

- Help them listen to their body. If they need a nap, let them take one without guilt.
- Schools may allow rest breaks during the day, if needed.



"At first, I felt like I had to do everything perfectly. Now, I focus on making each day the best it can be."

– Parent of a child with ARKPD

REVIEW: Key Points - Supporting Your Child and Yourself

Parenting a child with ARPKD or CHF is a journey. There will be hard days, but there will also be moments of joy and strength.

- **Lean on support groups and other parents**—you are not alone.
- **Advocate for your child's needs**, whether at school, in healthcare, or socially.
- **Celebrate small victories**—a good day, a new friend, or a successful doctor's visit.

Appendix A: Resources for More Help

ARPKD Database

arpkdb.org

- Children's Hospital of Philadelphia established a NIDDK-funded interdisciplinary center of excellence in PKD-related research, with specific emphasis on ARPKD.

Clinical Trials Information

www.clinicaltrials.gov

- This is a site managed by the U.S. National Institutes of Health (NIH) where you can learn about clinical studies from around the world.

go.pkdcure.org/ARPKDACTHandbook

- Sign up for the Accelerating Clinical Trials (ACT) Alerts to receive emails about ARPKD-specific clinical trials looking for participants.

Social Media Pages for ARPKD

PKD in Children (PKD Foundation)
facebook.com/groups/PKDinChildren

ARPKD
facebook.com/groups/ARPKD

ARPKD Angels
facebook.com/groups/arpkdangels

PKD Foundation Support Options

PKD Parents Community
pkdcure.org/get-connected/community/parents

- The PKD Parents Community has served parents of children with ARPKD and ADPKD since 2000. The PKD Parents Community offers support, compassion, and guidance, while helping connect families with experts and other PKD families.

PKD Thrive Community
pkdcure.org/for-parents/pkd-thrive

- Launched in 2022, the PKD Thrive Community brings together young adults with PKD. This community offers support, compassion, and a safe place to connect and interact with others who understand where you are in your PKD journey.

HOPE Line
pkdcure.org/get-connected/hope-line

- Our team is available Monday through Friday, from 8 a.m. – 5 p.m. (CT), to answer your questions and direct you to resources that will address your concerns.

PKD Connect Peer Mentors
pkdcure.org/get-connected/peermentors

- PKD Connect Peer Mentors provide resources, guidance, motivation, and emotional support to individuals impacted by PKD.

Grief and Loss Support

Now I Lay Me Down to Sleep
www.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.

Appendix B: What Families Should Know When a Baby Dies from ARPKD

Losing a baby is heartbreaking. If your baby had **autosomal recessive polycystic kidney disease (ARPKD)**, please know you're not alone. There are people and resources to support you through this incredibly painful time.

Why Some Babies Don't Survive

Even with the best medical care, about **1 in 5 babies diagnosed with severe ARPKD before birth** do not survive past infancy. This is often due to serious **breathing problems**, caused by underdeveloped lungs and large kidneys putting pressure on the chest.

Sometimes, parents and doctors face extremely difficult decisions about how much medical intervention to use or whether to focus on comfort care. These are deeply personal choices.

It's okay to ask for help—from your care team, from counselors, and from people who understand grief. Some hospitals have a grief and bereavement program that gives families support and helps coordinate care and resources while in the hospital and after. You can find additional support and bereavement resources on page 43.

Bereavement Photography

Some families choose to take **end-of-life or after-life photographs** with their baby. These are called bereavement photos, and they can become treasured memories. Professional photographers trained in this area can gently, and respectfully, help capture these moments. Many hospitals with neonatal intensive care units (NICUs) can help arrange this for you.

You don't have to decide right away, but it's okay to ask what is available.

Saving Tissue for Genetic Testing or Research

If your baby has died, you may be asked about **preserving tissue or blood samples**. These samples can help:

- Confirm the exact genetic cause of ARPKD
- Guide genetic counseling for future pregnancies
- Contribute to ARPKD research, which may help other families in the future

These are tough decisions to make in the middle of grief, so it may be helpful to talk with someone ahead of time if you know your baby's condition is very serious. **The PKD Foundation Parents Community** can help explain your options and connect you with the right people.

If You've Lost a Baby to ARPKD, Here Are Some Things to Consider:

- **You are not alone.** Connect with other bereaved parents. Often, your local hospital will be able to help you find local support groups and you can also seek support through the PKD Foundation Parents Community at **pkdcure.org/parents**.
- **Ask about support services** like palliative care, spiritual care, or bereavement specialists—they can be a lifeline. See bereavement resources on page 43.
- **You have the right to take your time** with decisions like photography, autopsy, or tissue donation.
- **You can honor your baby** in whatever way feels right—through memory boxes, naming ceremonies, donations, or simply sharing their story.
- **There are people who want to listen.** Whether now or later, you can always connect with the PKD community for guidance and support.



Appendix C: Understanding Tests to Monitor Your Child's Health with ARPKD/CHF

If your child has autosomal recessive polycystic kidney disease (ARPKD) or congenital hepatic fibrosis (CHF), their doctor will use different tests to monitor their **kidney, liver, and overall health**. These tests help track changes over time, detect problems early, and guide treatment decisions.

The most commonly used tests include **blood tests, urine tests, and imaging scans**. Your child's doctor may order additional tests depending on their specific health needs.

Blood Tests: Checking Organ Function and Overall Health

Blood tests provide **important information about kidney and liver function, blood health, and mineral levels**. These tests help doctors understand how well the kidneys and liver are working, detect complications, and decide if treatments or lifestyle changes are needed.

Kidney Function Blood Tests

Since **ARPKD affects kidney function**, doctors closely monitor how well the kidneys filter waste and maintain balance in the body. The following can all be done from a single blood draw.

- **Creatinine**: A waste product made by muscles that's filtered out by the kidneys. If the kidneys aren't working well, creatinine levels rise.
- **Cystatin C**: A newer marker of kidney function, which provides a more stable measure than creatinine because it's less affected by muscle mass. If the kidneys are not working well, cystatin C levels rise.
- **Estimated glomerular filtration rate (eGFR)**: A key estimate of kidney function, calculated using creatinine and, sometimes, cystatin C levels. Doctors use eGFR to help understand how well your child's kidneys are filtering waste.
- **Blood urea nitrogen (BUN)**: Another waste product from digesting protein. High BUN levels can indicate poor kidney function, dehydration, or a high-protein diet. It may be a less accurate reflection of kidney function than creatinine or cystatin C.
- Why it matters: **These tests help track how well your child's kidneys are working and detect early signs of kidney failure**. This allows doctors to adjust medications, recommend diet changes, or plan treatments, like dialysis, if needed.

Electrolyte Blood Tests

The kidneys help **balance essential minerals** in the body. Electrolyte levels can change if kidney function is impaired.

- **Sodium**: Some children with ARPKD have low sodium levels, which can cause **fatigue or weakness**.
- **Potassium**: High levels can affect heart function and need to be controlled with diet or medications.
- **Bicarbonate (carbon dioxide, CO2)**: Helps maintain the body's acid-base balance. Low levels may indicate a condition called metabolic acidosis, common in kidney disease.
- **Calcium and phosphorus**: Important for bone health. Kidney disease can cause calcium levels to drop and phosphorus levels to rise, leading to weak bones.
- Why it matters: Imbalanced electrolytes can cause serious health problems, including heart and bone issues. Regular monitoring helps ensure your child gets the right treatments, supplements, or diet changes to maintain balance.

Liver Function Blood Tests

Since **CHF affects the liver and bile ducts**, blood tests help doctors track liver health and identify potential complications.

- Liver function tests (like AST, ALT, bilirubin, or GGT) may be normal or slightly abnormal in children with ARPKD/CHF.
- **Vitamin D** and **vitamin K** levels may be low if the liver isn't absorbing nutrients properly.

Why it matters: **Monitoring liver health helps prevent complications** like vitamin deficiencies, bleeding problems, bone health, or portal hypertension (high blood pressure in the liver).

Blood Counts (CBC – Complete Blood Count)

This test checks different types of blood cells:

- **White blood cells (WBCs)**: Help fight infections.
- **Hemoglobin**: A protein found in red blood cells that carries oxygen. Low levels (anemia) can occur if someone has iron deficiency and/or if the kidneys aren't making enough erythropoietin (EPO), a hormone that stimulates red blood cell production.
- **Platelets**: Help the blood clot. Children with portal hypertension may have low platelet counts, increasing the risk of bleeding.

Why it matters: **Anemia and low platelets are common in children with ARPKD/CHF**. If needed, doctors may prescribe iron, EPO, or other treatments to improve blood health.

Bone Health Blood Tests

Two tests can help doctors understand whether your child might have problems with creating healthy bones.

- **Vitamin D level**: This vitamin helps the body absorb calcium and phosphate from food, which are necessary for healthy and strong bones.
- **Parathyroid hormone (PTH) level**: High levels of PTH cause bone loss because it signals the bones to release calcium into the bloodstream.

Why it matters: **Some children may require treatment with vitamin D supplements** if their vitamin D level is low and/or their PTH level is high.

Urine Tests: Checking for Kidney Damage and Infections

Urine tests provide valuable information about **kidney function and overall health**.

- **Proteinuria**: Protein in the urine can be a sign of kidney damage. If protein levels are too high, doctors may prescribe medications to reduce strain on the kidneys.
- **White blood cells (WBCs)**: If WBCs are present, it may indicate a urinary tract infection (UTI).
- **Urine culture**: Helps identify bacteria if a UTI is suspected.

Why it matters: **Kidney damage can progress silently**, so urine tests help catch problems early and guide treatment.

Imaging Tests: Getting a Clear Picture of the Kidneys and Liver

Imaging scans **allow doctors to see organ size, structure, and blood flow**, helping to detect complications.

Ultrasound (Most Common Imaging Test)

- Safe, painless, and radiation-free
- Checks for cysts in the kidneys and liver
- Measures kidney and liver size to track growth over time
- Helps detect portal hypertension by checking spleen size and blood flow

Why it matters: **Ultrasound helps doctors monitor disease progression** and plan treatments before serious complications arise.

MRI (Magnetic Resonance Imaging)

- **Magnetic resonance cholangiopancreatography (MRCP)**: Provides a detailed view of the liver and bile ducts.
- **MRI elastography**: Measures liver stiffness, which can indicate CHF progression.
- **Kidney MRI**: Less common but sometimes used in research studies.
- **No radiation**: Uses strong magnets instead.
- **Gadolinium contrast agent**: Sometimes used but is generally avoided in children with severely reduced kidney function.

Why it matters: **MRI helps doctors get a clearer view of liver and kidney disease** without using radiation.

Echocardiogram (Heart Ultrasound)

- Checks heart health in children with high blood pressure (hypertension).
- Can detect left ventricular hypertrophy (thickening of the heart muscle), which can happen due to chronic high blood pressure.

Why it matters: **High blood pressure can lead to serious heart problems**, and regular echocardiograms help doctors adjust treatment to protect heart health.

Why These Tests Matter

Children with ARPKD and CHF **need lifelong monitoring** to catch changes early and manage symptoms. These tests help doctors:

- **Track disease progression**: Is kidney or liver function getting worse?
- **Detect complications early**: Are there signs of high blood pressure, anemia, or electrolyte imbalances?
- **Adjust treatments**: Should medications, diet, or supplements be changed?
- **Plan for future care**: Is dialysis or a transplant needed in the future?

By staying on top of these tests, you and your child's medical team can **work together to keep them as healthy as possible**.

Appendix D: Caring for Your Family and Yourself After Diagnosis – Advice from an ARPKD Parent

When your child is diagnosed with ARPKD/CHF, your whole world shifts. You're suddenly balancing doctor's visits, missed work, and the challenges of keeping your child on track socially and in school. It's a lot. And while your focus is on your child, this diagnosis affects your whole family—including you.

I've learned that the right support can make all the difference. Hospitals, schools, family, and friends can help in ways you might not expect. The hardest part is often asking—but trust me, it's worth it.

Taking Care of Yourself as a Parent or Caregiver

When we first got our child's diagnosis, I went through all the emotions—grief, anger, fear. But over time, I shifted into **advocate mode**. That's what we do as parents, right?

The thing is, advocating for a child with a chronic illness is exhausting. It affects **everything**—health, emotions, school, relationships, family life. I had to learn (sometimes the hard way) that I cannot give my child my best if I **don't** take care of myself, too.

What Helped Me—And Might Help You

- **Ask for help—and accept it.** If someone offers to cook a meal, watch the kids, or run an errand, say yes. People **want** to help, but they may not know how unless you tell them.
- **Lower your standards (seriously).** It's okay if the house is messy or if you order takeout three nights in a row. Your energy is better spent on things that really matter.
- **Take time for yourself.** I used to feel guilty about this, but I've realized that self-care is not selfish—it's survival. Whether it's a walk, a book, a bath, or just sitting in silence, find something that refuels you.
- **Plan time with friends or your partner.** Even if it's just a quick coffee or a phone call, staying connected helps.
- **Do what makes you happy.** Some people love spa days; others find peace in organizing a closet. Whatever relaxes you—**do that**.
- **Have someone over.** Another adult in the house, even just for an hour, can give you the chance to **nap, bake, read, or just breathe**.

- **Divide and conquer.** If you have a partner, talk about who does what. You're in this together.
- **Let go of perfection.** I had to remind myself (often!) that I don't have to do it **all**. No one does.

A Final Thought

The people who love you want you to be okay. Your child needs you to be okay. And while this journey is overwhelming, you are not alone.

You've got this. And if today feels too hard, that's okay, too. Tomorrow is another chance.

"Connecting with other parents with children with PKD has been crucial for me. When my boys were first diagnosed, it was so helpful to have someone to talk to who had been through what I was going through. Helping families with newly diagnosed children has helped me feel like I am doing something positive for the disease.

Participating in fundraising efforts for PKD has helped me feel that I am making a difference and empowered me."

– Parent of a child with ARPKD

Appendix E: Going to School with ARPKD/CHF: Tips for Success

A chronic illness such as ARPKD/CHF can have significant impacts on a child's educational experience. These impacts may include missing school, sports, activities, and social events due to medical appointments or illnesses. It's 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.

Because ARPKD/CHF is such a variable condition, the degree of impact on children's education can differ greatly. Some children experience very few problems, whereas others may be more significantly impacted. For example, if they're receiving dialysis or a kidney transplant or are experiencing frequent illnesses. It's important for families to work with their medical team, social worker, and school to make sure the child's educational needs are being met. Some children may need special school accommodations, such as a **504 plan** or an **Individual Education Plan (IEP)**.

What is the Difference Between a 504 Plan and an IEP?

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

Children have different rights under IDEA (IEP, or Individual Education Plan) and Section 504 (504 Plans). An IEP is used when the disability impacts their ability to learn. Whereas a 504 plan is for children who may need accommodation due to their medical condition, but 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 mwedical 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	You are provided parents' rights in writing. There is a process for disputes. Services are free.	You 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 includes extended test time, reading tests to children, smaller workgroups, co-teachers or aides, plans on how to deal with difficult behavior, and homebound services.	• 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 the school year

Tips for 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 advocate all day in various ways for their health. That can lead to some conflicting 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 teaching experience and training. Be sure to begin your conversation with them by acknowledging their expertise.
- **Put concerns in writing**: Be prepared for meetings by providing a list of concerns. This can help minimize the number of meetings needed.
- **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's 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.

Appendix F: Supporting Your Child's Emotional Health with ARPKD

You are Not Alone

An ARPKD diagnosis is overwhelming—emotionally, physically, and mentally—for both the child and the entire family. There is no roadmap for how to help your child through the emotional ups and downs of this disease. But there is support and there are strategies that can help.

"My child didn't have the words to tell me how scared they were. They just started refusing to go to school. That was anxiety, not misbehavior."

Why Emotional Health Matters

ARPKD is not just a physical illness. It can deeply affect a child's emotional and mental well-being—sometimes even before they have the vocabulary to express how they're feeling. Kids might not say *I'm anxious* or *I'm depressed*. Instead, they may act out, withdraw, or get sick more often.

As a parent, your role is powerful. By learning what to look for and responding with compassion and care, you can be your child's strongest advocate.

Recognizing Anxiety in Children

Anxiety is more than nervousness. It can show up physically, emotionally, and behaviorally.

Physical signs of anxiety can include:

- Restlessness or being on edge
- Trouble sleeping
- Stomachaches, nausea, or "butterflies"
- Sweaty palms or racing heart
- Feeling dizzy or lightheaded
- Muscle tension or headaches

Behavioral signs might include:

- Refusing to go to school or leave the house
- Clinging to parents, pets, or safe objects
- Avoiding friends or activities they once enjoyed
- Self-harming behaviors, like picking skin, cutting, or hitting objects

Thought patterns in anxiety often sound like:

"What if something bad happens?"

"Everyone will laugh at me."

"I feel like I'm dying."

Understanding Depression in Children

Depression in kids does not always look like sadness. It often shows up as anger, irritability, or withdrawal. Two hallmarks of depression include feeling sad, empty, or hopeless for most of the day.

Physical signs of depression can look like:

- Loss of interest or pleasure in activities previously enjoyed
- Constant fatigue or feeling "weighed down"
- Aches, pains, or stomach issues
- Trouble sleeping (too much or too little)
- Changes in appetite or weight

Behavioral signs can look like:

- Expressing feeling sadness, emptiness, or hopelessness over several weeks
- Not caring about appearance or hygiene
- Quitting school, sports, or social activities
- Trouble concentrating or completing tasks
- Saying things like, *No one cares about me* or *The world would be better without me*

If your child talks about wanting to die or harm themselves, **contact your pediatric care provider or a qualified mental health professional immediately**. Do not wait.

What You Can Do

Listen First

Let your child share how they feel—even if it's hard to hear. Show them you can handle their feelings and they're not alone.

Use Creative Outlets

Kids may open up more through drawing, storytelling, music, or play. Follow their lead and get creative.

Find the Right Support

Look for:

- **Play therapy**: for younger children
- **Counseling or talk therapy**: for teens
- **Family therapy**: to support everyone

Respond quickly if they ask to speak with a therapist. That's a big, brave step.

Do not Forget the Siblings

Siblings of children with ARPKD often feel left out, jealous, or even guilty. They might not show it, but they notice when one child gets more attention.

- Consider **play therapy** for siblings, too.
- In families where multiple children have ARPKD, kids may support each other—but they still need space to process their own feelings.

Siblings need to feel seen, supported, and safe to express their own emotions—even when the focus is on a sick brother or sister.

Mental Health Is Health

Just like dialysis or a transplant requires expert care, so do mental health concerns. There's no one-size-fits-all solution, but with the right team, progress is possible.

Treatment options may include:

- Therapy or counseling
- Medication (in coordination with your child's kidney/liver team)
- Journaling or storybook creation
- Mindfulness, yoga, and gentle exercise

REVIEW: Key Points - Caring for Your Child's and Family's Mental Well-Being

Children with ARPKD are at higher risk for anxiety and depression, often shown through physical symptoms or behavior changes. Parents are powerful advocates—your calm, open presence makes a difference.

- **Listen, watch, and respond.** If something feels off, trust your instincts and seek help.
- **Mental health support** is important for siblings, too.
- **If there are any signs of suicidal thoughts** or self-harm, contact your pediatric care provider or a qualified mental health professional immediately.

Appendix G: Glossary of Terms

A

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

A condition where the body doesn't have enough red blood cells to carry enough oxygen to the tissues throughout the body. 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 20-50% of normal kidney function. Anemia tends to worsen as CKD progresses.

Autosomal recessive

Autosomal recessive is a pattern of inheritance. An autosomal recessive disorder occurs if an individual inherits two variants (changes) in a gene (one variant from each parent). The parents are carriers but don't have the disorder.

Autosomal dominant polycystic kidney disease (ADPKD)

ADPKD is one of the most common monogenic (caused by a mutation in a single gene) conditions. It is characterized by the development and growth of numerous fluid-filled cysts in the kidneys. The resulting damage frequently leads to kidney failure.

Autosomal recessive polycystic kidney disease (ARPKD)

ARPKD is a rare genetic condition, usually diagnosed in childhood, where multiple fluid-filled cysts form in the kidneys. While disease severity is highly variable, the growth of the cysts often causes loss of kidney function.

B

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.

Bile ducts

Small tubes that drain bile from the liver.

Blood pressure (BP)

A measurement of the force of the blood in the arteries 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's 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.

C

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 variant like that for ARPKD. They do not have the disease but can pass the variant 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 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)

A condition with 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 a 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 used as 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 used as 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).

D

Deceased donor

Most often individuals who die from accidents, heart attacks, or strokes, who's next of kin consent to donating their organs.

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.

E

Echocardiogram

Pictures of the heart taken using ultrasound.

Echogenic

When an organ, such as the kidneys or liver, appears 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.

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 bone marrow to make red blood cells. Individuals with abnormal kidney function may require a synthetic EPO supplement.

Estimated glomerular filtration rate (eGFR)

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.

F

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.

G

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 that 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.

H

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.

Hypertension

A condition when someone's blood pressure (the measurement of the force of the blood in the arteries as it flows through the body) is too high.

I

In vitro fertilization (IVF)

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

K

Kidney failure

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 (kidney function) is less than 15%. You may also see the term end-stage renal disease or ESRD for short. This is the same as kidney failure.

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.

L

Left ventricular hypertrophy

Thickening of the heart muscle in the main pumping chamber (left ventricle).

Liver function tests

Blood tests such as AST, ALT, bilirubin, and GGT, which help to assess different functions of the liver.

Living donor

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

M

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

A change or variant ("typo") in a person's genetic code.

N

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.

Neonatal intensive care unit (NICU)

Also known as an intensive care nursery, a NICU is an intensive care unit that specializes in the care of ill and/or premature newborn babies.

P

Paired donation

A transplant option for kidney transplant candidates who have a medically eligible living donor but cannot receive their kidney due to incompatibility (i.e., a poor match). In this process, the candidate and their donor are paired with another candidate and donor who also face incompatibility with each other, but whose blood types are compatible with theirs. The two recipients then 'swap' donors so each can receive a kidney that is a compatible match.

PD (peritoneal dialysis) 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's an injury to the body.

PKHD1

The gene that codes for ARPKD.

Poor kidney function

The gradual loss of kidney function. Your kidneys filter waste 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 waste 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

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.

R

Recessive

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

Red blood cells (RBCs)

Cells in the blood containing the red protein, hemoglobin, that are responsible for carrying oxygen throughout the body.

Rejection

A process where an individual's immune system "attacks" a transplanted organ (such as a kidney or liver).

S

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.

U

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).

V

Variant

A change or mutation ("error") in a person's genetic code.

Varices

Abnormally large (dilated) and sometimes twisted veins in the esophagus (tube that runs from the throat to the stomach) or the stomach. Over time, the walls of these veins can become weaker, allowing them to bulge and sometimes rupture and bleed.

Vascular access (hemodialysis)

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

Vitamin D

A vitamin that is important for bone health. It's 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.

W

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).

Notes





PKD FOUNDATION
Polycystic kidney disease

pkdcure.org
1.800.PKD.CURE