

# **PKD Patient's Manual**

## **Understanding & Living with Autosomal Dominant Polycystic Kidney Disease**

*By Irene Duley RN ANP & Patricia Gabow MD*

**PKD Foundation**  
Kansas City, Missouri

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## Dedication

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This booklet is dedicated to the many patients with polycystic kidney disease, their families and especially their children who daily displayed the courage that it takes to live with a chronic genetic disease. They truly are an inspiration to us all.

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## Mission

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The mission of the **PKD Foundation** is simple – “to promote research to find a cure for PKD and improve the care and treatment of those it affects.” A simple mission for a complex disease. All of the people connected with the **PKD Foundation** are dedicated to finding better treatments and a cure for this disease. The Scientific Advisory Committee, the **PKD Foundation** Board of Trustees, the staff of the **PKD Foundation** and most importantly, the people we serve, are looking for progress to be made in research so that we may keep our eyes focused on our vision that, “no one suffers the full effects of PKD.”

## **Introduction to ADPKD**

The purpose of this booklet is to provide information about Autosomal Dominant Polycystic Kidney Disease (ADPKD) to those who have the disease, those who are at risk due to an affected parent, invested family members and friends.

## **What is ADPKD?**

ADPKD is an inherited disorder. It is passed from one generation to the next by an affected parent to a child. Although the primary manifestation of ADPKD is cysts in the kidney, cysts as well as other abnormalities can occur in other areas of the body. The other organs that can be involved with ADPKD besides the kidney cysts are listed in Table 1.

## **How Common is ADPKD?**

ADPKD has been estimated to occur in approximately 1:400 to 1:1,000 live births. Although ADPKD has been seen throughout the world and among all racial and ethnic groups, it is not clear if the frequency is the same among all of these groups. Suffice it to say, ADPKD is common and affects 500,000-600,000 Americans. Imagine that every time you go to a local college football game, there is someone else in the crowd who has ADPKD like you.

### **Table 1**

Symptoms of ADPKD in adults may be some or many of these in addition to kidney cysts.

#### **Kidney**

Kidney cysts \*  
Enlarged kidneys  
Hypertension (High blood pressure)  
Back and/or side pain  
Blood in the urine  
Kidney stones  
Urinary tract infection  
Kidney failure

#### **Cardiovascular**

Mitral valve prolapse (floppy valve)  
Aneurysm (weakening and enlargement) of the aorta  
Left ventricular hypertrophy (Thickening of the heart muscle)

#### **Gastrointestinal**

Liver cyst formation  
Diverticula (outpouchings or weakened areas) in the colon

#### **Other**

Aneurysms (weakening and enlargement) of blood vessels to the brain  
Cysts in other organs, such as the pancreas or spleen (uncommon)  
Hernias

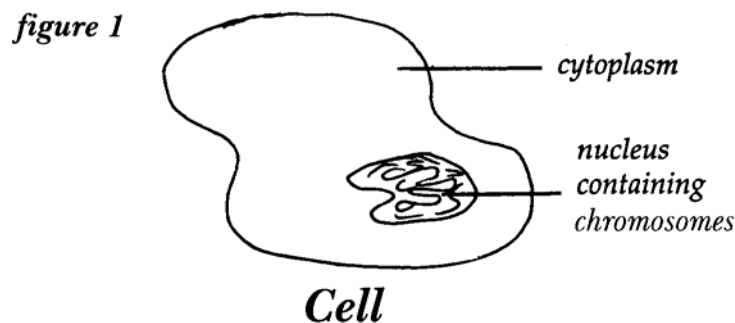
## How Does Inheritance Actually Work?

Our bodies are composed of billions of cells, all of which have two basic parts: the *nucleus* and the *cytoplasm* (fig.1). The *cytoplasm* is vital to proper cell function, while the *nucleus* is the control and operational center of the cell. It contains the message or *blueprint* inherited from our parents that determines what our cells actually do and what we will be like.

Each cell nucleus contains tiny threads called *chromosomes*. All the necessary information required to direct the formation and function of a human being is contained in these chromosomes. In turn, the chromosomes are composed of *genes* – the basic units of heredity. Genes are pieces of DNA so small they remain invisible even under an electron microscope. Therefore, genes are studied by molecular geneticists (is this last sentence really necessary – is the “molecular geneticists” thing ever mentioned again?)

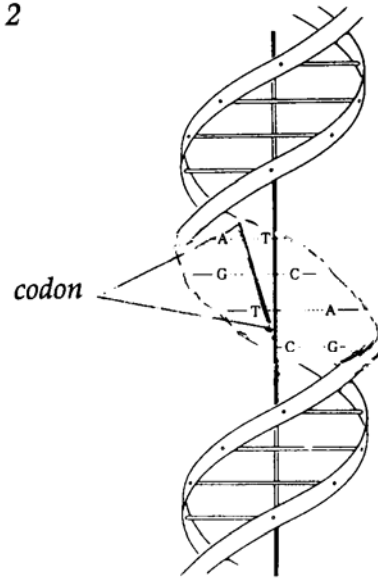
The building blocks of genes are chemical substances called nucleotides (fig. 2). There are four nucleotides: adenosine, thymidine, cytosine, and guanine, commonly expressed as A, T, C, and G. Ultimately, everything in our body depends on how these four nucleotides are arranged. A sequence of three nucleotides is called a *codon* (fig. 2) and genes are made up of codons. A small gene may contain 10 to 20 codons, whereas a large gene may contain many (do we have a number or general number we can put in here like, “thousands of codons,” or “millions of codons”?) codons. Each codon codes for a certain amino acid. In turn, these amino acids string together in a certain way to make a specific protein (fig. 3).

Each protein has a unique function in the body. In a disease that is genetically inherited, there is a *mistake* or *mutation* in the gene. A single nucleotide change is enough to cause the gene to code for an abnormal protein that causes a disease. Therefore, the goal in treating a genetically inherited disease is to find out what the abnormal protein does so that the usual function of that protein can be achieved. In the future, it may be possible to replace defective genes.



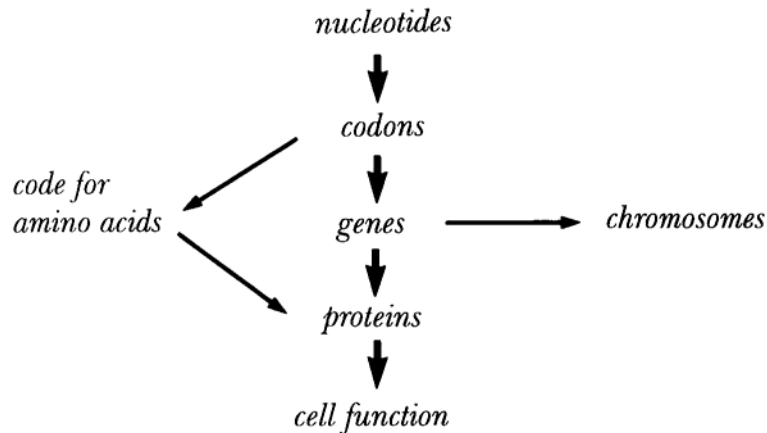
*Our bodies are made up of millions of cells. Genes in the chromosomes tell each cell exactly what to do.*

figure 2



*Deoxyribonucleic acid (DNA). A series of three nucleotides makes up one codon.*

*figure 3*



*Nucleotides are grouped to form codons; and codons are grouped to form genes. Each codon codes for a specific amino acid. The order of the codons determines the order in which the amino acids are grouped together to form a specific protein. Each protein in turn has a specific function in the body.*

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### **What Do We Know About the ADPKD Genes?**

There are two abnormal genes that cause ADPKD. About 80 – 85 percent of people affected with ADPKD have a mutation in the ADPKD gene located on chromosome 16, which is called *ADPKD1*. Virtually, the rest of the ADPKD population has the mutation in the ADPKD gene located on chromosome 4 called *ADPKD2* (fig. 4). The *ADPKD1* gene encodes the protein called *polycystin-1* while *ADPKD2* encodes *polycystin-2*. (NEW FIGURE) When these proteins function normally (not mutated), they are important in the regular (or “typical” or “standard” or “conventional”) development of kidney tissue.

It appears that the severity of the kidney disease caused by the *ADPKD1* and *ADPKD2* genes is slightly different. With the *ADPKD1* gene, cyst development, the onset of high blood pressure, and loss of kidney function appear earlier as compared to the *ADPKD2* gene, although there is a great deal of overlap.

Not so long ago, we knew almost nothing about the genetics of ADPKD. Now, not only do we know where two of the ADPKD genes are located, but the codons of both ADPKD genes have been determined and many mutations identified.

### How is ADPKD Inherited?

Every person has 23 pairs of *chromosomes*, making a total of 46 (fig. 4). Twenty-two pairs are called *autosomes*, and one pair is specifically devoted to determining the gender of an individual. Because the ADPKD genes are on an autosome, men and women have an equal chance of inheriting this disorder.

During reproductions, the chromosome pairs split in the formation of female eggs and male sperm. The woman donates 23 of her chromosomes to the baby and the man donates 23 in the sperm. In this way, when the egg is fertilized by the sperm, it will have the normal complement of chromosomes (fig. 5).

There are four possible ways the egg and the sperm of parents where one has ADPKD can combine. Two will contain the chromosome with the gene for ADPKD and two will not (fig. 6). Therefore, each child of a parent who has ADPKD has a 50 percent possibility of inheriting the affected chromosome.

*figure 4*



*Example of the normal amount of chromosomes arranged in 23 pairs. Twenty-two pairs are called autosomes and one pair determines sex. This person is female and has two XX chromosomes and no Y chromosome. Chromosome 16 has the ADPKD1 gene on it and chromosome 4 has the ADPKD2 gene on it.*

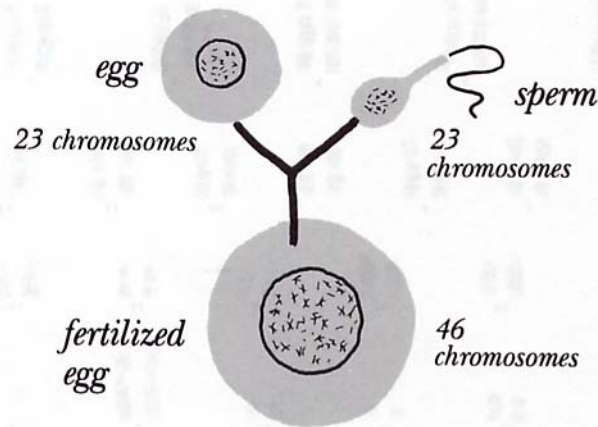
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**If I Have Four children, Does This mean That Two of My Children Will Have ADPKD and Two Will Not?** In real life, it may not work out that two children will have the disease and two

will not. The risk of having a child who inherits the chromosome with the affected gene is *always* 50 percent with each pregnancy, no matter how many children a couple has. It's like the flip of a coin – there is always a 50 percent chance of getting heads and a 50 percent chance of getting tails. In some families, all of the children are affected, while, in other families, none are. Many families with multiple children will have both affected and unaffected children.

In ADPKD, there is also approximately a 6 – 10 percent rate of *spontaneous mutations*. This means that instead of inheriting the ADPKD gene from a parent with the disease, the gene mutates by itself for no known reason. That is, the sequence of nucleotides or codons resulting in ADPKD has permanently changed. It is important to know that even with a true spontaneous mutation, a newly affected person will still pass the mutated gene on to his/her children.

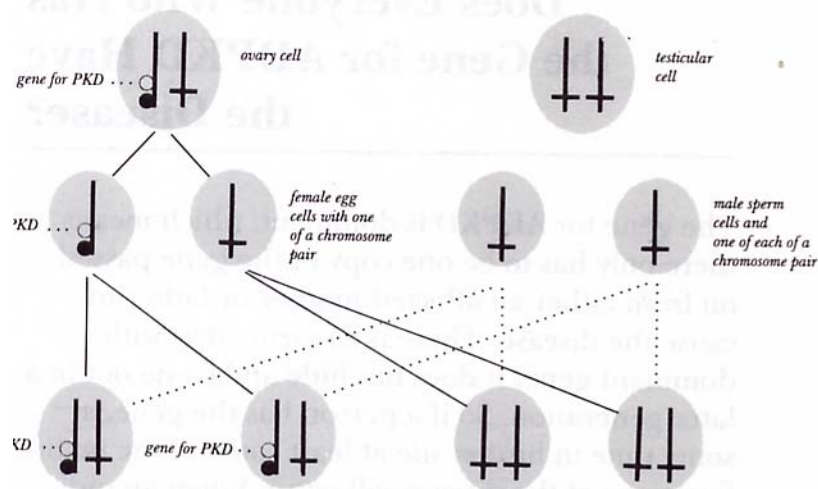
*figure 5*



*Female egg with 23 chromosomes (1 of each pair), sperm with 23 chromosomes (1 of each pair) and fertilized egg with 46 chromosomes (2 of each pair).*



figure 6



*Example of how PKD is inherited:*

*Two female eggs, one with the gene for PKD and one without.*

*Two unaffected male sperms.*

*Four possible combinations of egg and sperm fertilization.*

*The babies who inherit the PKD gene will have PKD. The babies who don't inherit the gene will not have PKD.*

### **Does Everyone Who Has the Gene for ADPKD Have the Disease?**

The gene for ADPKD is *dominant*, which means there only has to be one copy of the gene passed on from either an affected mother or father to cause the disease. There is no *carrier state* with a dominant gene – it does not hide and come out in a later generation. So, if a person has the gene, at sometime in his/her life at least some of the symptoms of the disease will occur.

**When an individual does not have the gene for ADPKD, he/she does not have the disease and therefore cannot pass the gene on to the next generation.**

However, that does not mean that everyone who gets the ADPKD gene has the same signs or symptoms, the same age of onset, or the same course of the disease. In fact, there is a wide spectrum of severity with ADPKD. At one end are children who are diagnosed before birth or in the first year of life with cysts and/or big kidneys, and at the other end is the person who has few, if any symptoms even when they are very old. Most people who have the ADPKD gene fall in the middle and at some time will have some of the signs or symptoms associated with ADPKD.

### **Does Everyone Who is in the Same Family Have the Same Type of ADPKD?**

Everyone does if the same family has the same type of ADPKD gene – ADPKD1 or ADPKD2 – and the same defect in that gene. However, even within the same family, signs and symptoms and the course of the disease are very often different. The most dramatic example of this occurs in families with children who are diagnosed before birth or in the first year of life. These

children have signs of the disease long before their parents with adult-onset PKD have signs and often the parents of children who were diagnosed before birth don't even know they have the disease until after their child is diagnosed and they themselves are subsequently tested.

Large ADPKD studies have also shown that kidney failure does not happen at the same age in members of the same family. In one large study, almost half of the families had a member who progressed to kidney failure more than 10 years earlier than did his/her parent. Because of this, it is very difficult to predict the course of the disease in any one family member by looking at the progression of the disease in his/her parent or siblings.

### **How Does a Person Found Out if He/She Has ADPKD?**

A physician is alerted to the possibility of ADPKD in three different settings:

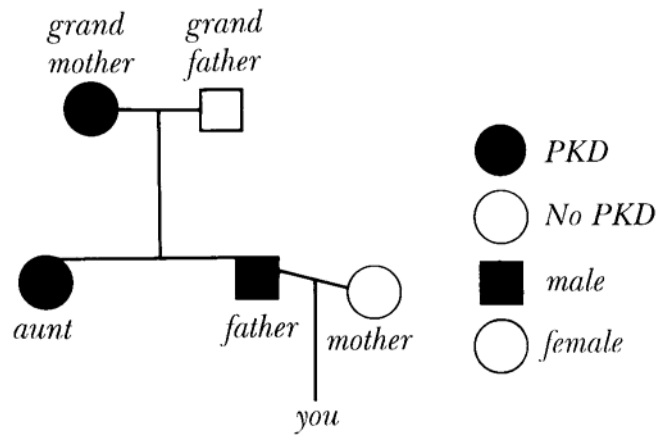
- When someone reports there is a family history of ADPKD,
- When there are signs and symptoms that commonly occur in ADPKD, or
- When a test is done for some other reason and cysts are found in the kidney.

About 70 percent of the time, there is a family history of ADPKD. Family history helps to see who is at-risk for developing cysts. In general, the signs and symptoms of ADPKD are not specific enough to permit a doctor to know if a person has the disease or not. For example, although some people with ADPKD have back pain and/or high blood pressure, so do many other people who do **NOT** have ADPKD. However, if a person goes to the doctor with these signs or symptoms and the doctor also feels enlarged kidneys or liver during an examination, he/she will likely think about ADPKD and order an ultrasound test.

*Ultrasound* is currently the most practical screening test for ADPK. Ultrasound can detect cysts in nearly all people with the ADPKD gene who are over age 30. It does not use dyes or radiation and is relatively inexpensive. Ultrasound may be negative prior to the age of 30 in 20 percent of patients who carry the ADPKD gene. For that reason it is important to remember that a negative ultrasound test before age 30 does not exclude the condition actually being present. *CT scans* or *magnetic resonance imaging (MRI)* can increase the sensitivity for diagnosis somewhat, but these tests are more expensive. The CT scan also uses x-ray dye which may potentially damage kidney tissue and both of these tests still have a false-negative rate of 15 percent before age 30. CT scans are sometimes used in patients with ADPKD to diagnose complications like bleeding into a cyst or the presence of kidney stones.

The limitations of all these methods are that the cysts have to be large enough to be seen. Many patients, particularly those with ADPKD2, do not have cysts large enough to see until they are 30 years of age or older. *Genetic testing* of a patient's DNA is available. This testing scans the individual's DNA isolated from a blood sample and looks for mutations in the PKD gene. However, this test is extremely expensive and mutations are found in only 60 – 70 percent of samples again giving us a high false-negative rate.

*figure 7*



*Example of an informative family for gene linkage analysis*

### **Since the Kidneys Are So Involved in ADPKD, What Should I Know About Them?**

Each person is born with two kidneys located in the back of the body on each side of the spine, tucked under the rib cage. The kidneys are about 5 ½ inches long (14 cm), 3 inches wide (8 cm), and 2 inches thick (5 cm) and they weigh 10 – 12 ounces (280 – 340 grams) (fig. 8). Both kidneys are affected when a person has ADPKD. There may be just a few cysts or many, and the cysts may range in size from a pinhead to a grapefruit. If there are many cysts, the kidneys can more than double in size and weight.

Each kidney contains about 1 million tiny tubes called *nephrons*. Almost a quarter of the blood your heart pumps every second is filtered through the filters in the nephrons called *glomerulus*. Red blood cells, white blood cells, and large substances like protein don't normally pass through the glomerulus, but rather stay in the body. The fluid that goes through the filter is made up of water, electrolytes and other small substances, and is about 180 liters (approx. 47 gallons) of fluid per day. This fluid passes through the glomerulus and the long tube of the nephron. All except 1 – 2 liters (a quart to a half-gallon) is reabsorbed back into the body; the rest ends up as urine. The process of filtering and reclaiming fluid along the nephron enables the normal kidney to perfectly maintain the body's fluid composition.

The kidneys are also a regulating system. They make sure your electrolytes such as sodium, potassium, calcium, phosphorus, and other chemicals are in balance and help regulate the pH of your bodily fluids so they are not too acidic or alkaline. The kidneys filter and excrete the waste products your body produces each day.

*Blood Urea Nitrogen (BUN)* and *creatinine* are two waste products removed by the kidneys. In particular, creatinine is removed so efficiently that an estimate of kidney function can be made by the level of this substance in the blood. Your doctor can calculate approximately how much actual kidney function you have with a blood test for creatinine, a 24-hour urine collection, along with your height and weight. This is called *creatinine clearance* and is approximately equal to

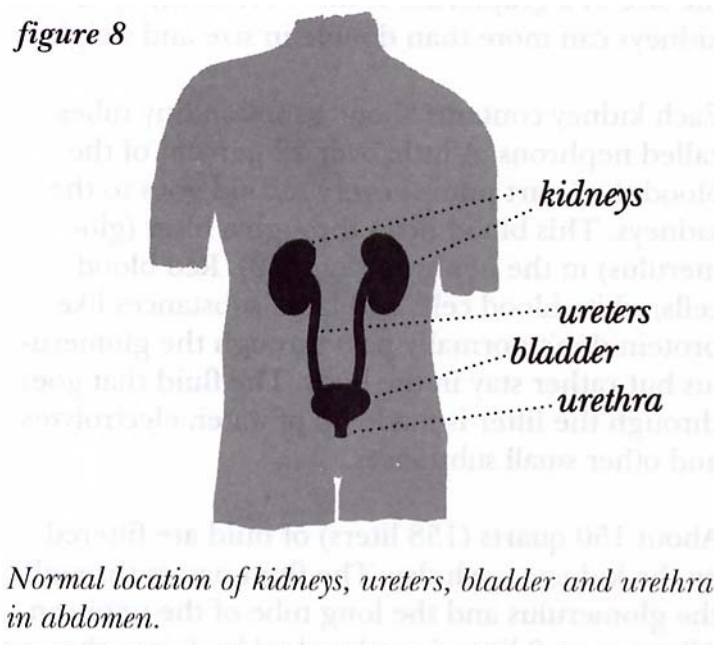
the true *glomerular filtration rate (GFR)* of your kidneys which can be measured precisely for research purposes. The creatinine clearance tells your doctor the approximate percent of “normal” kidney function you have.

The kidneys make several essential hormones. One of these is *renin*, an enzyme that facilitates the production of other hormones such as *angiotensin* (help regulate blood pressure) and *aldosterone* (aids in the body’s handling of salt and potassium). Another hormone made in the kidneys is *erythropoietin*, commonly known as *EPO*. This hormone tells the bone marrow to make red blood cells. If a person’s kidneys are surgically removed or if they fail because of a kidney disease, EPO is no longer produced and blood transfusions must be given to the person every five or six weeks. The exact gene that codes for the protein erythropoietin was discovered several years ago. There is now a genetically manufactured form of EPO that a person can take, which has all but eliminated the need for transfusions.

The kidneys are also required to change vitamin D to its active form, which helps the body absorb calcium from the diet. In this way, the kidneys help control calcium and bone formation.

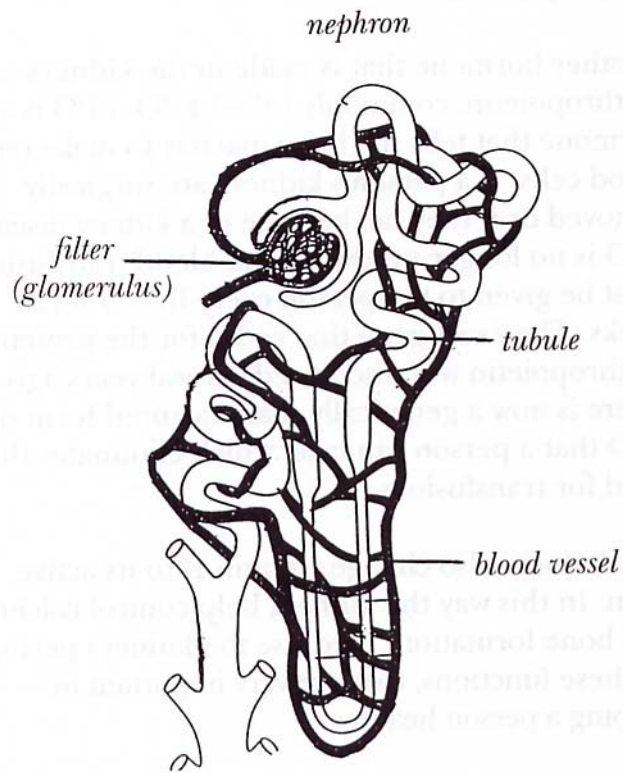
Because the kidneys perform all these essential functions, they are vital to keeping a person healthy.

*figure 8*



*Normal location of kidneys, ureters, bladder and urethra in abdomen.*

figure 9



*The nephron (kidney tubule) is the functional unit of the kidney. Substances and fluid pass from the blood vessel into the tubule. There are about one million nephrons in each kidney.*

### **What is a Cyst?**

A *cyst* in the kidney begins as an outpouching of the nephron, similar to a blister, and can occur anywhere along the length of the nephron. Although *polycystic* means *many cysts*, not every nephron forms cysts – only 5 percent of the nephrons do. The fluid inside the cysts often reflects the area in the nephron from which the cyst began.

Approximately 70 percent of cysts detach from the nephron when they are still very small, about 2 mm (1/8 inch) in diameter. Over time, the cysts enlarge and can become filled with clear fluid or fluid that contains blood or white blood cells.

Cysts can also form in other organs as well as the kidney, with the most common site being the liver. Current research suggests that liver cysts are associated with the bile ducts or tubules of the liver rather than liver cells themselves and that these cysts do not take the place of functioning liver cells, they merely push liver cells aside. This difference between kidney cysts

and liver cysts is the reason why liver cysts don't cause liver failure even though the liver can become quite enlarged due to cysts.

### **What Causes Cysts to Form?**

The genes that cause ADPKD give an incorrect message and make abnormal polycystins, which in turn, causes some cells in the body to function incorrectly. Research has shown there are at least three components to cyst formation (fig. 10):

- *Cell proliferation (growth)* – the cells of a cyst wall reproduce themselves more than do normal kidney cells, making them grow in size
- *Fluid secretion* – Secretion is a way of making fluid. To form a cyst, the cells themselves must secrete fluid. If there were no fluid produced to fill the cyst, there would merely be a ball of cells.
- *Abnormal basement membrane* – as its name implies, the basement membrane is a very thin layer of tissue the cystic cells sit on. In ADPKD, this layer is thicker than usual and is also incorrectly formed.

Research continues on other complex factors that affect cyst formation and growth.

### **How Do Cysts Cause Problems?**

In general, cysts cause problems because of their size and the space they occupy. Many of the symptoms people with ADPKD have are dependent on how large their liver and kidneys are. For example,

- people with kidneys over 15 cm (6 inches) are more likely to have pain than people with smaller kidneys,
- people with ADPKD who also have high blood pressure have bigger kidneys than those with normal blood pressure,
- people with ADPKD who have bleeding into their urine have bigger kidneys than those who don't, and
- people with large kidneys have more loss of renal function than people with smaller kidneys.

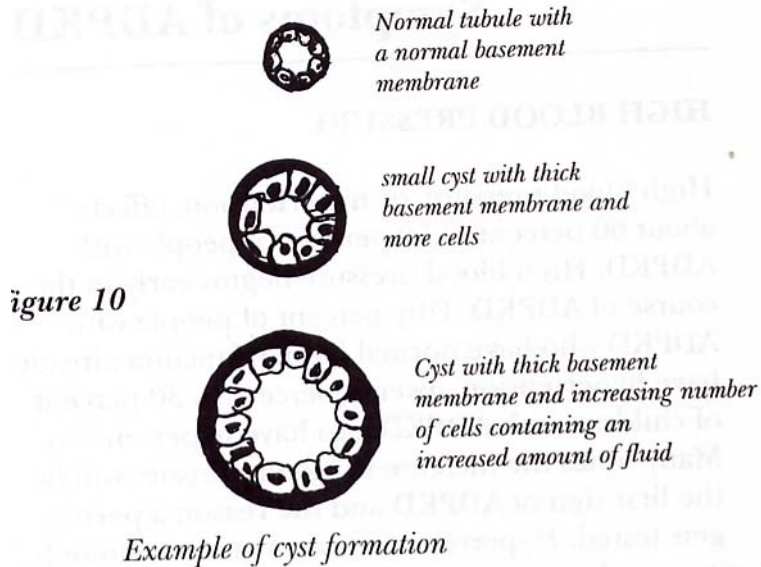


figure 10

### How Will I Feel If I Have ADPKD?

Early in the progression of the disease there are generally no symptoms at all. In fact, many people are never diagnosed with ADPKD because they either have no symptoms or a few symptoms that mimic other diseases. Often the first sign is high blood pressure, blood in the urine, or a feeling of heaviness or pain in the back, sides, or abdomen. Sometimes the first sign is a urinary tract infection and/or kidney stones.

### Can You Tell Me More About the Signs and Symptoms of ADPKD?

#### HIGH BLOOD PRESSURE (HYPERTENSION)

Blood pressure is a measurement of the force of the blood as it flows through the body. The pressure depends on the amount of blood and fluid in the body, how much blood the heart pumps each second (*cardiac output*), and the degree in which blood vessels are constricted or dilated. This is similar to the force it takes to get water through a garden hose. The pressure depends on how much water is going through the faucet and how narrow the hose is.

Blood pressure measurements have two parts, recorded as millimeters of mercury (mm Hg) – for example 120/80 mm/Hg. The top number or *systolic blood pressure* measures the pressure when the heart is actually pumping and the bottom number or *diastolic pressure* is the measurement of the pressure when the heart is relaxing between beats.

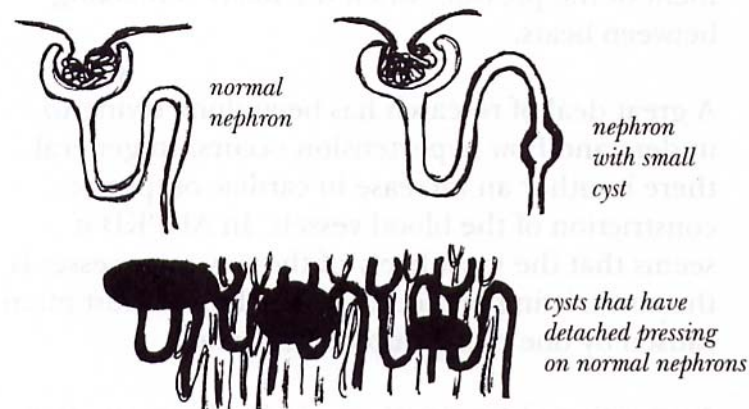
High blood pressure or hypertension affects about 60 – 70 percent of people with ADPKD and begins early in the course of the disease. Fully half of the people with ADPKD who have normal kidney function already have hypertension and is more common in men than in women. Twenty to thirty percent of children with ADPKD also have high blood pressure as compared to other children their age. Many times, the increase in blood pressure will be the first sign of ADPKD and is the primary reason a person gets tested. People with ADPKD and high blood pressure have more and larger cysts than do people with normal blood pressure.

A great deal of research has been done trying to understand how hypertension occurs. In general, there is either an increase in cardiac output or constriction of the blood vessels. In ADPKD, it seems the more likely of these two processes is the constricting of the blood vessels. This is thought to be related to excess activity of the renin-angiotensin-aldosterone system.

As mentioned previously, *renin* is a hormone produced in the kidneys and acts on *angiotensinogen*, a substance in the blood that forms yet another hormone called *angiotensin*. This hormone is a powerful constrictor of blood vessels and also stimulates the production of *aldosterone*, leading to salt retention and potassium loss.

In ordinary circumstances, the kidneys make renin when blood pressure is low and the kidneys sense they need a stronger flow of blood – in other words, a protective mechanism. In ADPKD, cysts can press on blood vessels in the kidney (fig. 11), resulting in decreased blood flow to some parts of the kidney. Sensors in the nephron react as though the blood pressure in the kidney was low, triggering the secretion of renin, which in turn generates angiotensin, constricting the blood vessels, and causing high blood pressure.

*figure 11*



*Nephron with no cyst, nephron with small cyst, and nephrons with many cysts.*

Whatever the initial cause of high blood pressure, it needs to be treated aggressively. If not treated, hypertension causes further damage to the kidneys, enlarges and thickens the heart muscle, and increases the risk for strokes and other cardiovascular events. Hypertension in ADPKD is often treated by a group of drugs called angiotensin converting enzyme inhibitors (ACE inhibitors) or angiotensin receptor blockers (ARBs). These two classes of drugs are usually the first drug of choice because of the role of angiotensin in high blood pressure in ADPKD. In general, both types of drugs are safe and effective, however, in some patients with large kidneys and decreased kidney function, these drugs can make kidney function rapidly worse.

Regardless of what kind of blood pressure medication is used, the most important thing is to have your blood pressure in or near the normal range, below 120/80 and the lower the better.



There are many different, very good medications to treat high blood pressure. Work with your doctor to find the right one for you. Remember, a blood pressure medicine only works if you take it, so try to have a regular time to take your medicine so that you don't forget.

Although drugs are important in treating blood pressure, in some individuals certain non-drug treatments can also lower blood pressure. Some of these non-drug therapies include weight loss, exercise, and a low-salt/high potassium diet. Even though these treatments have not been studied specifically in ADPKD patients, they are worth trying since they also improve a person's overall health.

**WEIGHT LOSS** Weight loss in some people who are overweight can lower or eliminate entirely the need for blood pressure medications.

**EXERCISE** In some people regular exercise can decrease blood pressure and improves muscle strength and heart function in everyone.

**ALCOHOL** People who drink more than three ounces of alcohol a day tend to have higher blood pressures than those who are nondrinkers or light drinkers.

**SALT** Even though salt intake has not been shown to be the cause of hypertension in ADPKD, a low salt diet may be helpful, particularly if certain drugs are being used.

**POTASSIUM** Although it has not been examined in ADPKD, in other causes of hypertension potassium can lower blood pressure. Therefore, a diet that is high in potassium is probably a good idea (see diet) **but only if your kidney function is in the normal range. Talk with your doctor before making any changes in your diet.** When kidneys start to fail they can't excrete the potassium properly, which could cause serious problems.

**CALCIUM/MAGNESIUM** In non-ADPKD settings, a deficiency of calcium and magnesium has been associated with high blood pressure. Dietary calcium and magnesium are best provided by dairy products with low-fat dairy products a good choice in maintaining normal mineral balance as part of healthy diet. Studies in high blood pressure patients without PKD have shown that the so-called DASH diet (Dietary Approach to Stopping Hypertension) which consists of lots of fruits and vegetables combined with low-fat dairy by itself lowers blood pressure substantially and a diet based on these guidelines would also seem appropriate for patients with ADPKD

**TOBACCO** Smoking increases the risk of heart disease and stroke, and when paired with hypertension these risks increase dramatically.

There is a close correlation between poor blood pressure control and progressive loss of kidney function with ADPKD. If you have ADPKD, you should have your own blood pressure cuff and monitor and keep a log of blood pressures away from the doctor's office. This will give your physician a better picture of your blood pressure over time.

## **Pain**

*Pain* can be *acute*, signaling a sudden new problem like bleeding into a cyst, infection or a kidney stone and it can also be a lingering problem, and then it is described as *chronic*.

*Chronic pain* is one of the most common problems for people with ADPKD. The pain is usually in the back or the side and occasionally in the stomach area. It can be intermittent and mild requiring only occasional mild pain medicine such as acetaminophen. However, in a small number of people, the pain can be constant and quite severe. For these few people, *cyst decompression* (drainage) can be very helpful. If there are a few big cysts that seem to be causing the pain, they can be emptied. This is done by using ultrasonography which allows the doctor to insert a needle into the cysts, drain the fluid, and then coat the cyst wall with alcohol so the cyst doesn't secrete more fluid. If there are a great many small cysts causing the pain, surgical decompression can be done.

Pain is a very subjective feeling. There is no way pain can be measured accurately except by the person who is experiencing it. It is also important to remember that pain frequency and tolerance vary greatly among individuals. Pain tolerance appears to be influenced by a person's cultural background, expectations, behaviors, and physical and emotional health. For this reason, pain clinics that utilize biofeedback and support groups can be very helpful to some people in managing their pain.

## **Blood in the Urine**

Close to 50 percent of those with ADPKD have had or will have *blood in their urine (hematuria)* at some point. The urine may look pink, red or brown. Passing small amounts of red blood cells in the urine that can only be seen under a microscope may also occur. This is called *microscopic hematuria*.

*Hematuria* is more common in an individual with large kidneys and high blood pressure. It is thought that the rupture of cysts or the small blood vessels around them is the cause, along with kidney and bladder infections and kidney stones as other probable sources of blood in the urine.

Blood in the urine can last for less than a day or the bleeding may go on for days. Notify your physician as soon as possible if you see blood in the urine. Strict bed rest, increased fluid intake, and acetaminophen (if there is pain) are the usual treatments if the bleeding is prolonged. Avoid taking nonsteroidal anti-inflammatory medications such as aspirin or ibuprofen as they may prolong the bleeding. If the bleeding is directly into a cyst, there may not be the presence of blood in the urine; however the pain can be severe.

There is a direct association between multiple episodes of hematuria and decreasing kidney function. Therefore, if a particular activity is generally associated with blood in the urine, it would be best to avoid that activity.

## **Urinary Tract Infection**

*Urinary tract infection*, commonly called *UTI*, is an infection caused by bacteria that have reached the bladder, kidneys or the cysts themselves. Other names used for UTI are *cystitis* for bladder infection and *pyelonephritis* when the infection is in the kidney but not a cyst.

The infection usually starts in the bladder when, if not treated, can progress up the ureters (the tubes from the kidneys to the bladder) into the kidneys. Although both men and women have UTIs, they are far more common in women, due to women having a very short urethra (the tube that goes from the bladder to the outside).

UTIs are quite common in the general population, but they may be more frequent in those with ADPKD. There is an association between frequent UTIs and worsening kidney function. Both males and females with ADPKD are more likely to have an infection after a catheter is placed into the bladder.

The most common symptom of UTI, particularly if the infection is in the bladder, is pain or burning with urination and/or an urgent need to urinate even though only a small amount of urine passed. When the infection is in the kidney or in a cyst, there may be a sudden onset of fever, chills and back or flank pain.

Your physician should be notified if any of these symptoms occur so that treatment can be started as soon as possible. Usually a urine sample will be taken for a culture to see what kidney of bacteria is causing the infection, and the appropriate antibiotic can be prescribed.

Women who have frequent bladder infections may decrease or eliminate the rate of recurrence by:

- Wiping from front to back after urinating or a bowel movement. This prevents dragging bacteria from the anus and vagina to the urethral opening.
- Drinking fluid prior to intercourse and urinating afterward. This may help flush out any bacteria that may have entered the urethra.
- For those who have frequent UTIs, antibiotics may be prescribed on a daily basis to prevent recurring infections.

### **Kidney Stones**

*Kidney stones* occur in about 20 – 30 percent of people who have ADPKD as compared to one to two percent of the general population. One reason kidney stones are more common may be due to cysts blocking the tubules, preventing normal drainage. When the urine stays in one area longer than it should, crystals can form and cause kidney stones. Uric acid and calcium oxalate are the two most common types of crystals that lead to stones. Another reason that stones may form in some people with ADPKD is that there is a decrease in *urine citrate*. Urine citrate is a substance that prevents formation of kidney stones.

The symptoms of kidney stones are severe pain in the back, side or into the groin. Often there will be blood in the urine when passing a kidney stone. Kidney stones are treated the same way they would if a person did not have ADPKD. Sometimes a machine using sound waves, called a lithotripter, may be used to break the stones into smaller pieces so they can be passed.

## **Does Everyone with ADPKD Eventually Need to Have Dialysis or a Transplant?**

Although everyone with the ADPKD gene develops kidney cysts, not everyone progresses to kidney failure, and if they do it's rarely before age 40. Several recent large studies have shown that about 70 percent of those with ADPKD have not progressed to kidney failure by age 50. These studies also showed that about 50 percent were not in kidney failure even at age 60, and 23 percent were not by age 70. There is a high probability that many people with a very mild form of ADPKD are unaware of their status and thus are never diagnosed.

Although we still don't know exactly how kidney failure happens in ADPKD, we do know some of the factors that increase the rate of progression to kidney failure. These include:

- Having the ADPKD1 gene as opposed to the ADPKD2 gene
- Male
- Diagnosed with cysts at a young age
- High blood pressure
- Large kidneys
- Multiple episodes of blood in the urine
- Urinary tract infections in men
- Being a woman with high blood pressure and four or more pregnancies

## **Are There Other Problems Associated with ADPKD?**

ADPKD is not just a kidney disorder; other organs can also be affected (table 1). The list that follows may look long and overwhelming, but it's important to remember that most people don't have all of these problems. If you have ADPKD, you and your family should be aware of the following possibilities so you can play a major role in your own care.

### **Liver Cysts**

Over ninety percent of people with ADPKD have cysts in the liver during their lifetime. *Liver cysts* occur in those under the age of 30 but they are small and often detectable only by sensitive MRI scanning.

The liver can remain normal in size with few cysts or become enlarged. Even though there is an increase in liver size, the amount of functional liver tissue remains fairly constant. Therefore, rather than have cysts take the place of normal tissue as occurs in the kidney, cysts in the liver seem to push good tissue aside. This appears to be the reason that normal liver function continues even with many cysts and enlarged liver size.

Liver cysts occur more often in women than men; women have liver cysts at a younger age and more and larger cysts than men. Women who have been pregnant are more likely to have liver cysts; and the cysts are also more numerous and larger in women who have been pregnant compared to women who have not been pregnant. This suggests that female hormones may influence the development of liver cysts. For this reason researchers are looking at the effect of estrogen in post-menopausal ADPKD women. Because estrogen may be a factor in liver cyst growth, the benefits of estrogen replacement therapy and the risk of liver cystic disease must be carefully weighed. A recent nurses' health study has disproved any benefit of estrogen to prevent cardiovascular disease, however estrogen replacement therapy is protective against osteoporosis and decreases vasomotor instability – a cause of hot flashes in post-menopausal women. Thus,

the quandary is to balance increases in the size of the liver cysts against the benefits of estrogen for post-menopausal symptoms and osteoporosis. Work with your doctor in determining what would be the best course for you.

Women with ADPKD who do make the decision to take estrogen after menopause should have a baseline ultrasound of their liver before they start estrogen replacement therapy (ERT) and every two years thereafter. This will help your doctor evaluate if liver cysts are increasing in number and/or size. It is unclear at this time if it is better to take ERT in pill form or by skin patch, but theoretically, the patch would be a better choice since oral therapy provides high concentrations of estrogen directly to the liver.

There have been reports of cysts in the liver becoming infected, with symptoms of liver cyst infection ranging from fever to pain in the abdomen on the upper right side. These symptoms need to be reported to your doctor as soon as possible. Treatment of an infected liver cyst usually requires drainage and antibiotic therapy.

Although not common, if the liver becomes much enlarged and pain is disabling, decompression of cysts may be indicated. When there are a few large cysts, they can be drained by inserting a needle through the skin. When there are more cysts and greater liver involvement, a surgical procedure can be performed to “unroof” the cysts and/or take a section of the liver itself out – a major procedure that is rarely required. If it is needed, surgery should be done by a physician skilled in the procedure since it can be accompanied by complications.

### **Mitral Valve Prolapse (MVP)**

*Mitral valve prolapse (MVP)* is a condition where the valve separating the top and the bottom of the left side of the heart does not close properly. Sometimes this causes blood to leak back to the top part of the heart. This is called regurgitation and can be heard during an examination of the heart as a heart murmur. Symptoms that can be associated with MVP are palpitations, a feeling that the heart is running away or that there are extra beats in the heart, and chest pain that is not associated with exercise or exertion. MVP occurs in approximately 26 percent of the people who have ADPKD compared to 2 – 3 percent of the general population. The majority of people with MVP never experience any major problems.

MVP is usually confirmed with an ultrasound of the heart valves called an echocardiogram. If MVP is present and causing palpitations that are bothersome, they can be treated with medications that can also treat high blood pressure at the same time. Stopping the use of caffeine, alcohol and cigarettes may be enough to decrease or stop the palpitations in many cases.

Rarely, an infection of a heart valve can occur as a complication of MVP. Although not a common occurrence, it can lead to destruction of the heart valve. Therefore, if you have MVP and a heart murmur, inform all physicians who care for you. To protect your heart valve from infection, antibiotics prior to certain surgical or dental procedures may be prescribed.

### **Intracranial Aneurysms**

An *aneurysm* is an outpouching in a blood vessel, which can leak or rupture. In these events the symptoms can include sudden severe headache, pain in moving the neck, nausea and vomiting,

difficulties with speech or movement, and even loss of consciousness. All such symptoms require *immediate* medical attention.

*Intracranial aneurysms* occur in the blood vessels of the brain (fig. 12). Recent studies done in the United States have shown that people with ADPKD have about a 5 – 10 percent risk of developing intracranial aneurysms. They also seem to cluster in certain families – that is, if a member of your family has an aneurysm or has ruptured an aneurysm, you may be at a higher risk of having an aneurysm yourself.

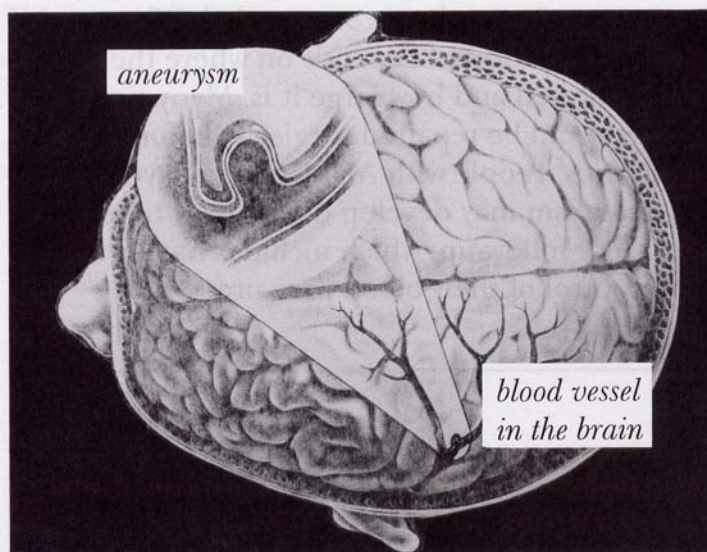
Because the risk for aneurysm is small, not everyone with ADPKD needs to be tested. However, people who have ADPKD and a family history of aneurysm *should* be tested, along with those whose job or hobbies would put them or others at risk if they lost consciousness (such as those who fly airplanes or drive buses). It's important to inform your physician if you have a family history of intracranial aneurysms and/or if you have a high-risk occupation or hobby.

*Magnetic resonance arteriography (MRA)* is usually the first test done to screen for cerebral aneurysm, while some centers prefer *high-resolution CT Scans*. Both of these tests are relatively noninvasive.

If there are any areas where an aneurysm is suspected, a *cerebral arteriogram* is usually performed. This test is more invasive and is done by putting dye directly into the blood vessels and will more clearly show if there is an aneurysm and how large it is.

If an aneurysm is found in someone with ADPKD, surgical repair or a therapeutic coil (a device placed in the aneurysm by a neuroradiologist) may be recommended. When and if surgery is performed depends on where the aneurysm is located and how large it is. Many times an aneurysm can be repaired surgically before it leaks or ruptures. People with ADPKD who have had one aneurysm may develop others over time and need periodic follow-up. Recent studies from the University of Colorado and the Mayo Clinic suggest a period of 5-10 years between follow-ups for aneurysms <10 mm in size, rather than annually. Blood pressure control is vital in these patients.

figure 14



*Blood vessels in the brain with an aneurysm.*

### **Hernias**

Both *inguinal* and *umbilical hernias* are more common in those with ADPKD. *Inguinal hernias* are outpouchings in the area of the groin and *umbilical hernias* are outpouchings at or near the navel. These should be surgically repaired if they are large or are causing other problems, just as they would be in someone who does not have ADPKD.

### **Diverticula**

*Diverticula* are outpouchings on the large intestine (colon). It seems that people with ADPKD who are on dialysis or have had a transplant have diverticula more often and also have more complications from diverticula, including infection, than people who have other kidney diseases. At the present time, we are not recommending any routine evaluation for this possible symptom.

### **Pregnancy: Can I Safely Have Children if I Have ADPKD?**

The diagnosis of ADPKD is no longer only made years after a person has had a family. The use of noninvasive ultrasonography has made the testing for ADPKD safe for all ages. Thus, screening specifically for ADPKD or finding evidence of ADPKD while doing an ultrasound for other reasons has made early diagnosis a more common occurrence. As the age of diagnosis is lower, an understanding of the risks of pregnancy in women with ADPKD is important.

The fertility rates of both men and women who have ADPKD are no different than they are for the general population. However, when you factor in end-stage renal disease and dialysis, there is a loss of both libido and fertility. Although most women do not get pregnant while on dialysis, they are not sterile. If a patient has a transplant, the libido more often returns to what it was prior to the loss of kidney function.

Generally, women with ADPKD who have normal blood pressure and normal kidney function have uneventful pregnancies and deliver healthy babies. Risk factors associated with pregnancy

and ADPKD are due to increased blood pressure. Some women with ADPKD will develop hypertension during their pregnancy and are more likely to have continued elevations in their blood pressure after delivery. Women who have high blood pressure prior to becoming pregnant have the risk of further elevations in their blood pressure while pregnant and women with complications in their first pregnancy are more likely to have complications in future pregnancies.

It is important for a woman with ADPKD to be closely monitored during pregnancy whether she has hypertension or not. Increases in blood pressure as well as protein in the urine could herald a serious complication of pregnancy called pre-eclampsia – a condition where the placenta can be prevented from getting enough blood. If the placenta doesn't get enough blood, the baby gets less air and food, which can cause low birth weight, premature birth, and other problems for the baby. Most women with preeclampsia still deliver healthy babies.

Pregnancy does not seem to affect the growth of cysts in the kidney. But there appears to be a mild long-term loss of kidney function in women who have hypertension and have had four or more pregnancies, compared to ADPKD women with hypertension who have fewer than four pregnancies.

The decision to have children is a very personal one. Both parents need to discuss both the risks involved and the joys associated with having a child. With an affected parent, there is a 50 percent probability of having a child who has inherited the gene for ADPKD (see “Genetics” above). All alternatives and risks, including attitudes on termination of pregnancy, should be addressed with the couple.

### **Is There a Special Diet That Will Make My Kidneys Better or Keep Them from Getting Worse?**

At this time, there is no specific diet that will make polycystic kidneys better or keep them from getting worse. However, one of the functions of the kidney is to remove waste products from the body. The major source of these waste products is the food we eat, especially protein. When a great deal of kidney function is lost, these waste products back up in the blood and cause the symptoms of kidney failure. Therefore, when a person has lost a significant amount of kidney function, a low protein diet may be ordered by his/her physician.

Some studies done on both animals and humans have shown that eating large amounts of protein at one time may cause kidney function to become worse more rapidly than eating smaller amounts of protein. The Modification in Diet in Renal Disease (MDRD) study done by the National Institutes of Health on protein intake and kidney function did not show any benefit from lowering protein intake in individuals with ADPKD. However, the people studied already had a significant loss of kidney function and future studies need to be conducted.

**Should I stop eating salt?** High blood pressure in ADPKD does not seem to be caused by salt intake, however, excessive amounts of salt should be avoided. This becomes important when people are on certain types of blood pressure medicine and when they have kidney failure.



**Can I drink alcohol?** Light and/or occasional use of alcohol has not been shown to damage kidneys or the liver. However, drinking three or more ounces of alcohol a day has been associated with increases in blood pressure and can damage the liver.

**Should I take extra vitamins or make sure that I'm getting all the nutrients I need?** A person who eats a fairly regular diet usually does not need extra vitamins. Unlike food, vitamins are needed only in tiny amounts. Excess amounts of vitamins A, D and E can accumulate in the body and cause medical problems. Generally, if you feel you need extra vitamins, a one-a-day generic brand of vitamin is sufficient. Consult your physician before taking extra vitamins of any kind. Because there is an increase incidence of calcium kidney stones in individuals with ADPKD, women with ADPKD should discuss with their doctor the proper amount of calcium needed. Low calcium diets do not prevent kidney stones in non-PKD patients and the beneficial effects of dairy product intake on skeletal and cardiovascular systems have already been established.

**How much fluid should I drink each day?** The body is usually very efficient in regulating the amount of fluid you need each day. You should drink when you are thirsty. Although the kidneys of people with ADPKD may not reabsorb fluid as efficiently as those without ADPKD, thirst is still the best way to tell how much fluid to drink.

**Will caffeine damage my kidneys?** There is no direct evidence that caffeine damages kidneys in people with ADPKD, however, studies done in test tubes combining ADPKD kidney cysts and caffeine-like substances have resulted in cyst growth. There is no clear relationship between caffeine and cysts in people since our bodies do not need caffeine (some may not agree with this, especially in the morning), it may be best to limit caffeine intake to 200 – 250 mg a day or two – three cups of coffee

#### **CAFFEINE CONTENT OF BEVERAGES**

<b>Beverages</b>	<b>Serving Size</b>	<b>Caffeine (mg)</b>
Coffee, drip	5 oz	110-250
Coffee, perk	5 oz	60-125
Coffee, instant	5 oz	40-105
Coffee, decaf	5 oz	2-5
Tea, 5-minute steep	5 oz	40-100
Tea, 3-minute steep	5 oz	20-50
Hot cocoa	5 oz	2-10
Coca-cola	12 oz	45

#### **Foods High in Potassium**

Potassium is essential to all living cells and is important for muscle and nerve functions in the body. It is found in most foods, such as legumes, whole grains, fruits, green vegetables, potatoes, meats, milk and yogurt. Although potassium is vital to the body, it is not wise to take potassium supplements in pill or liquid form without consulting your physician.

### **Can Children Also Have ADPKD?**

It was once thought that people who inherited the gene for ADPKD did not form cysts or have symptoms until well into adult life. We now know that ADPKD can be diagnosed at a very young age and even before birth. Any diagnostic tests done during pregnancy should be done in conjunction with medical counseling so the results of the test can be completely understood. For example, knowing a fetus carries the ADPKD gene does not predict the course of the disease. The fetus could go on to develop ADPKD in childhood or never have a symptom until much later in life.

There seem to be two different groups of children with ADPKD – those who are diagnosed before birth or in the first year of life with large kidneys and/or cysts, and those who are diagnosed after the age of 1.

Children who are diagnosed **in the first year of life** have some special characteristics:

- Most often the parent with ADPKD is the mother
- Most of these children have brothers and/or sisters who are also diagnosed in the first year of life.
- Most are diagnosed before birth with large kidneys, but often they do not have actual cysts.
- Most develop high blood pressure in childhood, so this should be watched for and treated.

Children who are diagnosed *after* 1 year of age are:

- Just as likely to have an affected father as an affected mother
- Cysts, even though their kidneys are not necessarily enlarged
- Affected with only one cyst in half the children, or just a few cysts. Whereas, in an adult one cyst alone is not enough to diagnose ADPKD because, as a natural part of aging, people often develop a few kidney cysts without having ADPKD. But in children who are a part of an ADPKD family, even one cyst means they are more likely to have the disease.

The number of cysts a child has affects his/her signs and symptoms. Just as in adults, children with many cysts are more likely to have back, side or stomach pain and are also more likely to have high blood pressure than children with only a few cysts.

Almost all children who are diagnosed after the first year of life have perfectly normal kidney function that seems to stay normal throughout childhood. Most children will maintain normal kidney function until they are into their mid-twenties.

### **What Kind of Medical Treatment Should a Child with ADPKD Have?**

As with adults with ADPKD, blood pressure should be measured regularly – at least every six months – in children with the disease. In children, normal blood pressure is different for different ages and it also varies between boys and girls. All PKD children who have high blood pressure require treatment and should be seen by a children's kidney specialist (*pediatric nephrologist*).

Although less common than in adults, signs and symptoms of infection, blood in the urine and/or pain also need to be evaluated by a physician.

### **Should I Limit the Physical Activity of a Child Who Has ADPKD?**

There is no information to support limiting physical activity in any child simply because he or she has ADPKD. It is possible that children with large kidneys and/or large cysts may have more episodes of blood in the urine if they play contact sports such as football, however, each child should be evaluated on an individual basis.

### **Should I Tell My Children They Have or Are at Risk of ADPKD?**

To date, no research has been done on the effect such knowledge would have on children. Generally speaking, there is no need to burden children with information they are too young to understand. Children have a tendency to ask questions when situations arise and, at that time, children usually want simple honest answers. There is no need to go into great detail unless a child asks more questions on the subject.

Children of affected parents need not be tested for ADPKD. They may be monitored by their physicians for blood pressure, urinalysis and general health without actually making the ADPKD diagnosis. The decision parents make to test a child should recognize the fact that a negative result in childhood may not exclude the diagnosis later in life. The added consequences of making the diagnosis in childhood may give the child a label which will result in discrimination in employment and possibly in health and life insurance. Children can be informed of their risk for ADPKD but routine screening is not recommended at this time. When and if therapies become available to prevent the progression of cystic kidney disease then the decision to screen may change.

### **Do Children with ADPKD Have Involvement of Organs Besides the Kidney?**

Just as in adults, children who have ADPKD are more likely to have mitral valve prolapse (MVP) and hernias than children without ADPKD. Approximately 12 percent of all ADPKD children will have MVP, but unlike adults, it is unusual for them to have any symptoms. When children do have hernias, they should be treated as they would in any other child. Children rarely have any of the other manifestations of ADPKD.

### **What Should I Do to Better Take Care of Myself if I Have ADPKD?**

- Find a doctor who you trust and with whom you work well.
- Be involved in your own health care and become your own expert by gathering as much information as possible about ADPKD and any other health problems you may have. This will assist you in knowing your choices and allow you to make well-informed decisions. Pay attention to symptoms and write them down, including details such as when symptoms started, what time of day they occur, how long they last, what makes them better or worse to give your physician a clearer picture when you discuss your concerns with him or her. Ask questions and make certain you understand your care
- Know about the medications you are taking. When your doctor prescribes a drug, don't be afraid to ask questions like:
  - What are the advantages of this drug?

- What are the possible side effects?
- Is it dangerous to take this drug with any foods, beverage or other medications I'm taking (including over-the-counter medications)?
- Will any other condition I have be aggravated by this drug?
- Are there alternatives to this drug (generic brand, other medication, different treatment)?

Ask your pharmacist questions regarding over-the-counter medications and your medical condition. **NEVER** take medications that were prescribed for a friend or other family member.

- Exercise on a regular basis. Regular exercise improves stamina, decreases stress, enhances a sense of well-being, may have a beneficial effect on blood pressure, and especially with aerobic exercise, better cardiovascular function. It was once thought that people with kidney disease were unable to participate in an active lifestyle. Since then, many studies have shown that in kidney patients, exercise may be even more important than in the general population. People do much better on dialysis and with a transplant when they are physically fit.

Generally, people with ADPKD can do any activity they want unless they get blood in the urine or the exercise causes back, flank or abdominal pain. The exercises that are least jarring to the kidneys are walking, swimming and biking. The key is to find an activity that is comfortable for you and that you enjoy doing.

### **How Can I Tell if My Kidneys Have Failed?**

*End-stage renal disease (ESRD)* is when normal kidney function declines and needs to be replaced by dialysis or transplant. At this point kidneys can no longer balance electrolytes and acids in the blood or remove the wastes and excess water. These imbalances will result in your not feeling as well as you are used to. The progression to end-stage renal failure is usually gradual for people with ADPKD.

Symptoms that some people experience during this time are:

- Decreased energy
- Weakness
- Shortness of breath
- Weight loss
- Nausea and/or vomiting
- Metallic taste in the mouth
- Mild to moderate depression
- Decreased ability to think problems through.

It's important to keep your doctor informed of your symptoms so she/he can help you decide when it's time to start therapy. Blood tests will show that your blood urea nitrogen (BUN) and creatinine are not being eliminated by the kidneys and are building up in the blood. These tests may also show that your electrolytes and pH are out of balance.

In many centers, planning for kidney replacement therapy (*dialysis*) is done when there is about 25 percent of kidney function still left. If a person waits until he/she is very sick (*uremic*), it will take much longer to recover and may require more and longer hospitalizations.

### **What Options Are There for Me if My Kidneys Fail?**

The types of treatment for kidney failure include *hemodialysis*, *peritoneal dialysis*, and *kidney transplantation*.

**Hemodialysis** – a procedure that removes extra fluid, electrolytes and wastes using a dialysis machine. This can be done in three different settings.

- *Home hemodialysis* – dialysis that is done at home with an assistant and your own dialysis machine
- *In-center, self-care hemodialysis* – dialysis done in a center with you doing as much as possible with the assistance of staff at the dialysis center
- *In-center hemodialysis* – dialysis that is done in a center with the staff providing all of the care.

**Peritoneal dialysis** – a type of dialysis that removes extra fluid, electrolytes and wastes using the lining of the abdominal cavity (peritoneum). There are two ways to do peritoneal dialysis.

- *Continuous ambulatory peritoneal dialysis (CAPD)* – dialysis done on a continuous basis with exchanges four times a day
- *Continuous cyclic peritoneal dialysis (CCPD)* – dialysis done during the night using a machine to make the exchanges while you sleep.

The basic principle of dialysis is to have the blood flow on one side of a natural or artificial membrane, with special fluid on the other side. This membrane permits molecules that have built up in the blood to pass into the special fluid and be removed.

### **How Will I Choose Between These Types of Treatment?**

When the time comes that you need dialysis, your physician and the dialysis team will discuss in detail all the options available to you. Often when a person gets close to needing dialysis, they take a tour of the dialysis facilities, where they can talk to others on dialysis and the nursing staff to get a sense of what works best for them and their family.

Kidney transplantation is the treatment of choice for most patients with ADPKD. With transplantation, a healthy kidney is placed in the lower abdomen and takes over the function of the failed kidneys. The types of transplant are:

- ***Living related donor*** – when a member of your family donates a kidney
- ***Living unrelated donor*** – when a more distant relative, spouse or friend donates a kidney
- ***Deceased donor*** – when a kidney comes from a donor who has recently become brain dead but whose kidneys are functioning normally.

### **Since ADPKD is a Hereditary Disorder, Can Family Members Be Kidney Donors?**

A family member can be a kidney donor if that family member does not also have ADPKD. The first step for a potential donor is to have an ultrasound made of his or her kidney. Eighty-three – 90 percent of people at risk for inheriting ADPKD can be diagnosed with ultrasound by age 30.

If the family member doesn't have ADPKD, the transplant team can continue the evaluation to make sure everything else is acceptable for the person to donate a kidney.

### **Will My Kidneys Be Removed Before I Have a Transplant?**

Usually the kidneys are not removed before a kidney transplant is performed, however they will be if:

- There have been many infections
- There is bleeding
- There is a tumor in the kidneys.

Sometimes, one or both kidneys are removed if they are so large there is no room for the new kidney, or if the person is very uncomfortable or having problems eating because of kidney size.

### **How Are the Costs Associated with Dialysis and Transplant Covered?**

In general, Medicare covers a significant amount of the cost of dialysis and transplantation. To be eligible, a person must have earned Social Security benefits or be the spouse or dependent of someone who has. About 93 percent of those with end-stage kidney disease are eligible. For more detailed information regarding Medicare and payment of costs associated with dialysis and transplant, call your local Social Security Medicare office or write to the U.S. Department of Health and Human Services (address in back of booklet) for the Medicare Handbook. Another good source of information for Medicare coverage and kidney replacement therapy is your local transplant unit and The National Kidney Foundation also published a booklet on these issues. Those who do not qualify for Medicare may be covered by Medicaid for the indigent. A social worker or financial counselor in the dialysis unit or transplant program will help you work through the financial issues.

### **Common Tests Done When a Person Has ADPKD**

#### **BLOOD TESTS**

**Creatinine** is a measure of kidney function. It is a waste product of muscle metabolism (the work the muscles do). After creatinine leaves the muscles, it enters into the blood, then is filtered by the kidneys and ends up in the urine. There is always some creatinine in the blood and some in the urine. When there is a loss in kidney function, the kidneys do not clear creatinine from the blood as efficiently as they once did. This causes an increase of creatinine in the blood, which can be measured by a simple blood test.

**Normal blood creatinine is 0.6 to 1.4 mg/dl.** When a person's blood creatinine goes up to 2.0 mg/dl, they have lost approximately half of their kidney function.

**Blood urea nitrogen (BUN)** is another measure of kidney function. Urea nitrogen is the waste product of dietary protein. If there is a loss of kidney function, the urea nitrogen builds up in the blood. A number of factors such as diet, protein intake, heart function, and fluid status can affect BUN. Therefore, creatinine is the preferred measure of kidney function. The normal range for BUN is 6 to 15 mg/dl.

**Liver function tests** fall under tests on the blood. Liver function is almost always normal even if there are cysts in the liver. If at some time your liver function tests are not in the normal range, your physician should look for a cause other than ADPKD.

## **URINE TESTS**

**White blood cells (WBC)** in urine. WBCs normally are present only in small numbers in the urine. Some people with ADPKD do pass a few more, however, large numbers of WBCs in the urine suggest a urinary tract infection. If this happens, your doctor will culture your urine to determine if and what types of bacteria are present and from those results, plan a course of action.

**Red blood cells (RBC)** in the urine. Only a few RBCs are normally found in the urine. As with white blood cells, some people with ADPKD also pass a few RBCs. Sometimes with an episode of bleeding, there are so many RBCs that they color the urine pink, red or brown (see *hematuria*).

**Protein** in the urine. Protein is normally found in the urine only in small amounts. About one-third of those with ADPKD pass protein into the urine, but it is usually less than a gram in 24 hours. If protein loss is greater than one gram in 24 hours, there may be another problem occurring in the kidney along with ADPKD.

**24-hour urine collection.** This test is done in combination with the blood creatinine test to determine kidney function, called *creatinine clearance* or *glomerular filtration rate (GFR)*.

**Imaging studies.** Studies used to see or image organs or blood vessels in the body.

**Ultrasonography** is a test done with sound waves. Ultrasonography does not require the use of radiation or contrast dye to be injected into a person. Because it's so safe and accurate, ultrasonography is the most common imaging test done to screen for ADPKD and can be done safely in pregnant women..

**Echocardiogram** is an ultrasound of the heart. One of the uses of an echocardiogram is to image the valves of the heart. Your physician may order this test if he or she suspects you have mitral valve prolapse (MVP).

**Computed Axial Tomography (CT Scan)** is a sophisticated form of x-ray. CT scan uses radiation and, most often, contrast dye to visualize the organ or blood vessels being studied. This imaging technique is very helpful if complications of ADPKD arise, such as bleeding into a cyst or kidneys stones. *High-resolution CT scan* is used to visualize the blood vessels in the brain. This type of CT can show if there are any suspicious areas that may be aneurysms before doing more invasive or expensive tests.

**Magnetic Resonance Imaging (MRI)** takes pictures of the body using a magnet that puts a certain spin on particles that exist in a person's body and does not require contrast dyes or radiation. Cysts are easily visualized with MRI and appear much better than ultrasonography in following the course of ADPKD.

**Magnetic Resonance Arteriogram (MRA)** is used to visualize the blood vessels in the brain for screening of aneurysms. This is similar to high-resolution CT, but does not use contrast dyes or radiation. MRA is used when people are allergic to contrast dyes and/or iodine, or if they have lost kidney function.

**Arteriograms** are procedures that utilize contrast dye injected into the blood vessels in order to clearly visualize them. An arteriogram is usually called for when it is suspected that there is an aneurysm on a blood vessel in the brain.



## **Resources:**

### **For Further Information:**

American Society of Nephrology  
1725 I Street, NW, Suite 510  
Washington, DC 20006  
[www.asn-online.org](http://www.asn-online.org)

American Society of Transplantation  
17000 Commerce Parkway, Suite C  
Mt. Laurel, New Jersey 08054  
[www.a-s-t.org](http://www.a-s-t.org)

Medicare: Coverage of Kidney Dialysis and Kidney Transplant Services  
Medicare Handbook (CMS Publication #10128)  
U.S. Department of Health and Human Services  
[www.medicare.gov](http://www.medicare.gov)

National Kidney Foundation  
30 East 33<sup>rd</sup> Street  
New York, New York 10016  
[www.kidney.org](http://www.kidney.org)

PKD Foundation  
9221 Ward Parkway, Suite 400  
Kansas City, Missouri 64114-3367  
[www.pkdcure.org](http://www.pkdcure.org)

## **Articles**

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