

THE FAMILY AND ADPKD:

A Guide For Children and Parents



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General Information

What is ADPKD?

ADPKD is an inherited or genetic disease that affects many different parts of the body, but most notably the kidney.

What are kidneys and how do they work?

Everyone has two kidneys located on either side of the backbone, under the skin and muscles and the small of the back (see Figure 1). In general, each person's kidney is about the size of their fist. Each kidney contains 1 million working units called nephrons (see Figure 2). An individual nephron begins with a filter, called glomerulus, that is connected to a long tubule. The glomerulus is made up of a small coiled blood vessel and a thin membrane. Water, salts and waste products filter from the blood, across the membrane, into the tubule and eventually leave the kidney as urine.

The kidneys provide several important functions to maintain a healthy body: 1) they regulate the amount of water and salts in the body; 2) they rid the body of excess acid and waste products; 3) they help control blood pressure; 4) they play an important role in maintaining proper levels of calcium and other building blocks for bones; and 5) they produce hormones that are important in making new blood cells and in maintaining healthy bones.

What are polycystic kidneys?

Polycystic means "many cysts." Cysts are fluid-filled balloons. In ADPKD, cysts can form in many places in the body and in the kidney, anywhere along the nephron (see Figure 3). This process of cyst formation almost always involves both kidneys. The fluid in the cysts resembles urine made by the kidneys. These cysts grow and push hard on the good parts of the kidney. The growing and pushing of these cysts causes two problems: 1) the kidneys grow very big, and 2) the good kidney parts are damaged and eventually are lost because of the constant squeezing. In some but not all people, the loss of the good kidney parts causes the kidneys to stop working. Usually this does not happen in children, but it can happen in adults age 40 years or older.

Are only the kidneys affected in ADPKD?

No, ADPKD affects many different parts of the body. However, the kidney involvement is one of the most important problems in children. Other organs such as the liver, heart, and blood vessels can become affected in adults with PKD.

Inheritance

How did I get ADPKD?

ADPKD is a genetic disease. In this type of genetic disease, one of your parents has the abnormal ADPKD gene and has passed it on to you. Just like you, your parent with ADPKD got the disease gene from one of their parents.

Every person has 25,000-35,000 genes and there are two copies of each of these genes. Only one copy of the ADPKD gene is needed to cause the disease. So, a person with ADPKD has one normal copy and one disease copy.

Why did I get ADPKD and some of my brothers and sisters did not?

A baby receives one copy of each gene from their mother and one copy from their father. Which gene copy each parent gives the baby is decided entirely by chance, sort of like the flip of a coin.

In diseases like ADPKD, only one abnormal copy of the gene is needed to cause the disease. In ADPKD families, each baby has a 50% chance of inheriting the ADPKD gene from the affected parent. For each new baby, the risk is the same. It doesn't matter whether any other child in the family has PKD. For example, there are some families with 11 children and none of them have ADPKD. There are families with four children and all of them have ADPKD. The affected parent has no control over which copy of the ADPKD gene is passed along to his or her children. Figure 4 gives an example of how ADPKD gene can be passed down from one generation to the next.

Diagnosis

I feel fine. What's the point of finding out whether I have ADPKD?

No one wants to find out that there is something "wrong" with them or that they have a disease. It is true that many children who inherit the ADPKD gene have no problems during childhood. But other children with ADPKD do develop medical problems and many of these problems may be present without the child realizing that anything is wrong. Sometimes, children from ADPKD families are diagnosed with ADPKD after doctors notice their blood pressure is too high. High blood pressure is common in ADPKD, and it can be without symptoms, or silent. That is, there isn't anything about high blood pressure that warns someone they have it. Other times, ADPKD is diagnosed after blood is found in the urine. It usually takes a lot of blood in the urine to change its color. However, doctors can test urine with special color sticks to tell if even a small amount of blood is present. For patients with ADPKD it is important to treat high blood pressure as well as blood in the urine. Therefore, your doctor should check your blood pressure and your urine at every visit. If any abnormalities are found, you and your parents, along with your physician, should consider kidney tests to see if you have ADPKD.

Sometimes children from ADPKD families develop infections in their kidneys or aches and pains in their backs, sides or bellies. If it hurts when you urinate or if you have any of these sorts of pains, it is important to tell your parents, your teacher or any other important adult who helps to take care of you. The cause of the infection or pain may be ADPKD.

In addition to being able to treat medical problems that may be caused by ADPKD, once your doctors knows you have the disease, she or he can be on the lookout for other important and treatable conditions associated with ADPKD. In addition, if you know that you have ADPKD, you and your family can pay extra attention to your diet and to activities that are potentially risky for your kidneys.

For just about everyone who inherits the ADPKD gene, the day will come when a doctors makes the diagnosis of ADPKD. News of having a disease that will be with you for your entire life is upsetting. But it is important to remember that the disease gene has been present since birth. Finding out that you have ADPKD does not suddenly make you sick. Even so, many people who are at risk for inheriting the ADPKD gene do not want to know whether they have it. This is completely understandable. It is not necessary to find out particularly if your blood pressure remains normal, your urine is free of blood and you don't have any pain. On the other hand, if you have any of these problems, finding out can give everyone the best chance of preventing ADPKD-related complications from happening.

Am I different from other kids because I have ADPKD?

No, not really. Until about 10 years ago, it was very unusual to find children with ADPKD, probably because it is a disease that changes slowly over decades, rather than years. Most affected people found out that they had ADPKD when they were 30 or 40 years old. Something made them go to the doctor. Either they developed pain in their back or bellies, or they saw blood in their urine, or they had a kidney infection.

Sometimes they went to the doctor for their yearly checkup and their blood pressure was noticed to be very high. For these adults, kidney and heart damage due to ADPKD is usually already present and this damage is not reversible.

Today, children are diagnosed with ADPKD for one of two reasons. Some children develop very similar kinds of medical problems as adults with the disease. But, far more commonly, families with ADPKD now understand how the disease gene can be passed on to their children and they want to know if their children have ADPKD in hopes of preventing kidney damage. Being diagnosed with ADPKD simply means that you have a kidney disease that may cause problems later in life. Most children with ADPKD feel perfectly fine and do not have medical problems during childhood.

Taking Care

Is there anything I can do about having ADPKD?

Because ADPKD is a genetic disease, at this point people have PKD for life. Many scientists are trying to understand the ADPKD gene in hopes of finding a cure. Until this happens, though, the best that we can do is to help prevent cysts from growing and minimize further damage to the kidney. There are lots of good habits that can prevent further damage to your kidneys.

First, avoiding dehydration is very helpful. In ADPKD, the kidneys can have trouble holding onto water. Therefore, it is important to bring enough water and liquids with you on long hikes, bike trips and camping trips. Likewise, if you play sports like soccer, gymnastics or basketball, it is important to drink lots of liquid so as to avoid becoming dehydrated.

Second, avoiding large amounts of red meat can help protect your kidneys. The kidney works hardest when it has to clear waste products. Large amounts of red meat make kidneys work very hard. It is probably a good idea to eat a hamburger or a small steak only a couple times a week and to include other good sources of protein in your diet, such as chicken, fish, beans, and pasta.

Third, if you start to feel sick, particularly if you have a fever or pain when you urinate, it is important to tell your parents or your teacher or whoever takes care of you, immediately. We have learned that the earlier we find a kidney problem, like an infection, or bleeding, or breakage of a cyst, the more easily we can limit the amount of kidney damage.

Is there anything I shouldn't eat or drink because I have ADPKD?

While it is important for all children to have a healthy and balanced diet, it is particularly important for children with ADPKD. Too much salt in the diet can increase blood pressure. We worry about high blood pressure in people with ADPKD because it can cause faster loss of kidney function than if the blood pressure is normal. High blood pressure also damages the heart. Just about all foods contain some salt and some salt in the diet is O.K. What you should avoid is too much salt in your diet. This may be difficult because you may not always have control over what you eat and you may like the taste of salt. But, minimizing the amount of salt that you eat is one of the easiest ways to control your blood pressure. The first place to start is to never use a salt shaker at the table. Some foods contain a lot of salt and should be kept to a minimum. These are canned soups, pickles, potato chips, french fries, processed meats (like hot dogs) and many frozen dinners. Of course, "fast food" such as hamburgers and pizza usually contain large amounts of salt. A good rule of thumb to use is, the faster a prepared food can go from its container into your mouth, the more salt it contains.

Red meat is another issue to consider. A big red meat meal contains lots of protein. After you eat a large amount of protein, the kidney must work especially hard to clear the waste products. Any kidney that is damaged should not be forced to work too hard. But protein is important particularly for growing children. This is where balance is very helpful. It isn't necessary or wise to take all meat out of your diet, but it is probably best that you not eat a huge steak

every few days. We have learned a lot about protein and the work that dietary protein forces the kidney to do. We know that certain protein-rich foods like fish, chicken, turkey and beans are easier on your kidneys than steak or roast beef. We also know that small portions of red meat several times a day are easier on your kidneys than one huge steak. So there are ways to balance things in your favor while you can still enjoy the flavor!

Are there any sports that I can't do because I have ADPKD?

There are no hard and fast rules about limiting sports for children with ADPKD. However, experiences taught us that contact sports, such as competitive football, rugby, hockey, and basketball, can place children with ADPKD at increased risk for popping or tearing a cyst. When that happens, you will have pain in your back or sides and/or bloody urine. Also, sports that involve repetitive bouncing, such as horseback riding, may increase the risk of rupturing or popping a cyst. On the other hand, ADPKD doesn't limit the level to which you can push yourself physically, as long as you do not have pain or bloody urine. There are ADPKD patients who are world-class cyclists, runners and swimmers. Clearly, excelling in sports is not out of the question in people with ADPKD.

The most important issue when you participate in sports is to be sure to stay well hydrated. Playing any sport for a few hours without drinking lots of liquids can be dangerous to anyone with ADPKD. In addition, if during or after playing sports you notice blood in your urine, or pain in your side or back, it is important to let your parents or your coach know right away.

I see my mother or my father taking pills. Does this mean they are sick?

No, not necessarily. Remember the expression that an ounce of prevention is worth a pound of cure? In many cases, people take pills to prevent bad things from happening to them. Just as people without PKD may take vitamins or iron tablets to make their bodies healthier, ADPKD patients may take medication to lower their blood pressure or to slow the rate at which the cysts are growing. This doesn't mean they are sick. As a matter of fact, almost 40 million Americans without ADPKD take medication every day to control their blood pressure. If someone in your family is taking medication every day, they are doing the right thing to keep themselves as healthy as possible.

Sometimes people with ADPKD do get sick and need different or stronger medication for a period of time. In many ways, this situation is not very different from someone getting the flu or a bad cold. Very rarely, people with ADPKD develop a more serious problem and they need to be admitted into the hospital. The good news is that doctors have learned a lot about ADPKD and its associated problems, so patients shouldn't be sick very often or for very long.

I don't have high blood pressure. If I have to take a pill, does that mean I am sick?

No, not necessarily. In children with ADPKD, the kidney cysts can grow quite a bit, even without the child feeling any different. Everyone with ADPKD is different. In some children, the cysts can grow quickly, while in other children the cysts may not grow at all. It is not yet clear what makes cysts grow. However, we are learning that there are ways to

stop cysts from growing or from increasing in number. Currently these ways are being tested in clinical studies. Therefore, even if you don't feel sick, or have any discomfort, or even if you don't have high blood pressure, your doctor who is involved in such a study may give you a pill to help stop cysts from growing in your kidneys. In fact, this may become the most common and, in the long run, most important reason for children with ADPKD to take medicine.

Some of my family (mother, father, aunt, uncle, grandparents) have been very sick and died because of ADPKD. Is this going to happen to me?

No, this won't necessarily happen. No one has a crystal ball to predict the future, but we know so much more today about ADPKD than we did even five years ago. Therefore, many of the problems that other people had in the past can be treated or even prevented today.

For example, many years ago, we didn't have as many good antibiotics to fight kidney cyst infections. We didn't have as many medicines to lower blood pressure. Many people didn't find out that they had ADPKD until their kidneys stopped working and they needed dialysis. Years ago, there weren't as many doctors who could care for sick patients with ADPKD. Many times, patients had to wait a long time to see a doctor, and important kidney function was lost while they waited. Finally, dialysis and kidney transplantation only started in the 1970s, and until the last 20-30 years, it was not uncommon for people to die of kidney failure. This is very uncommon today.

We aren't saying that ADPKD can no longer make you sick. But, you should be reassured that most people with ADPKD can lead a healthy, active and full life. Doctors know a lot more about ADPKD and about how to minimize your chances of getting sick.

One of my aunts is on dialysis. What happens with dialysis and kidney transplantation? If I need a kidney transplant, how long will it last?

In general, patients receive dialysis when their kidney function has decreased to less than 10 % of normal. Dialysis treatments remove waste products and excess water from the body. There are two forms of dialysis: 1) hemodialysis, in which waste products are removed from the body by circulating blood through a special machine, and 2) peritoneal dialysis, in which waste products are removed into a special fluid that has been put into the belly cavity by a tube. However, dialysis cannot completely replace all of the functions of a normal kidney. Some of the important hormones and other factors that the kidney produces need to be taken by pill or injection.

Because dialysis does not work quite as well as a normal kidney, transplantation is usually a better long term treatment than dialysis. Experience has shown us that ADPKD patients generally do well following kidney transplantation. Unfortunately, we cannot precisely predict how long transplanted kidneys will work. Many transplanted kidneys have worked well for 10-20 years or even longer, but others have stopped working sooner. With current medications to suppress rejection (the process by which the body tends to fight the transplanted kidney), 75%-80% of transplanted kidneys work adequately for at least five years. There are many new drugs being developed, and it is hoped that these new medications will help to keep the transplanted kidney functioning for many years.

My uncle donated a kidney to my father who had ADPKD. My brother has ADPKD too. How can I decide whether to donate a kidney to my brother?

Kidney transplants provide the opportunity for someone who has lost their kidney function to lead a more normal life and avoid dialysis. Transplanted kidneys from a healthy family member usually last longer than kidneys obtained from people who have died and donated their organs. Donating a kidney is a very special act, but it may not be the right thing for everyone to do. Potential kidney donors must be chosen very carefully to be sure that they will not have any medical problems living with just one kidney. In addition, in ADPKD families, doctors must be very sure that the kidney donor doesn't also have ADPKD.

Donating a kidney is more than just giving a kidney away. It involves a very personal exchange with another person. Doctors take great care to be sure people are really ready to give away a kidney before they begin the process of evaluating someone as a suitable donor. No one is ever forced to donate a kidney.

FOR PARENTS AND OTHER ADULTS

This section is for you because you have a child, grandchild, niece, nephew or other child for whom you deeply care who either has ADPKD or who is “at risk” for having ADPKD.

Clearly no adult wants to see children sick or carry the risk of developing a disease, particularly if that disease has been passed on from either you or your spouse.

However, as a parent or concerned adult, one thing that you can do is to share your experiences and your wisdom, so that your child can be more familiar with ADPKD and less afraid.

Diagnosis and Prognosis

Our children are at risk for ADPKD. Given the potential problems with medical insurance, we are reluctant to have them screened for PKD. What do you suggest?

The decision to screen children is a very personal one. In part, it depends on which devil you can live with, the devil of knowing or the devil of not knowing. In making this decision, you also have to weigh the pros and cons. A "pro" is the possibility of avoiding or treating complications such as high blood pressure, blood in the urine or infection, if you and your doctor know that your child has the disease. On the other hand, cost of medical insurance could be prohibitive and obtaining life insurance could be difficult if your child is known to have a pre-existing medical condition. But if you don't know, you and your doctor are potentially without valuable medical information.

For example, we do know that kidney cysts grow and enlarge for a long time before there are symptoms or any loss of kidney function. Researchers are getting closer to finding therapies that could potentially slow down the rate of cyst growth in ADPKD, and these therapies could potentially help your affected child. When that day arrives, we hope in the not-too-distant future, it would benefit a child with ADPKD to find out they have the disease.

If you choose not to have your children screened for ADPKD, the safest thing to do is to inform your doctor that one of you has ADPKD and that your children might have the disease. With that information, your physician can then appropriately screen your children for treatable complications of ADPKD, particularly for high blood pressure. Given the changing climate in health care in this country, the difficulty in getting reasonable medical insurance at an affordable cost for someone with a pre-existing medical condition may soon improve dramatically. Your insurance agent may be able to help in planning for your child's long-term insurance needs.

What are the ethical guidelines for disclosing information regarding the diagnosis of ADPKD in a child?

Ethicists advise that if a child requests a diagnostic test, he or she should receive the results and be allowed to keep these results confidential, even from his or her parents. If the child refuses to disclose his or her diagnosis and there are risks for serious complications, the physicians can override this confidentiality agreement.

If you, the parent, requests testing, you should receive the results. If your child is old enough to have agreed to the testing, the results should be shared with your child. If small children are tested and found to be affected, they should have access to the test information by the time they reach adulthood, if they indicate a desire to know. It is the responsibility of the child's physician to carry out this wish.

I have a child who was diagnosed shortly after birth with ADPKD. I have heard that children diagnosed during pregnancy or shortly after birth will always need dialysis. Is that true?

Not necessarily. It is very rare that a child will need dialysis because of ADPKD. However, children who are diagnosed during pregnancy (in utero) or during the first year of life appear to have a more serious course with ADPKD than children or adults diagnosed later in life. These early-onset children often have high blood pressure earlier in life and have more cysts in their kidneys than children who are diagnosed later.

When the first early-onset ADPKD children were reported, they were very sick and in need of aggressive medical attention. Not uncommonly, these children were initially misdiagnosed with a more serious kidney condition called autosomal recessive polycystic kidney disease (ARPKD). These children were examined and diagnosed because they were sick. Now ultrasounds are routinely performed during pregnancies, and ultrasound machines are getting better and better in detecting tiny cysts. Therefore milder and milder forms of early-onset ADPKD are being found in babies and young children. In fact, most of these children have normal kidney function into adulthood and may have high blood pressure as their only medical problem during childhood. Whether these early-onset ADPKD children are different from other children who are diagnosed later in life is not clear. Scientists are actively studying this issue.

I have had both of my children undergo ultrasound screening for ADPKD at the ages of 7 and 9. Their ultrasounds are normal. Does this mean they do not have ADPKD?

Unfortunately, no. Children who carry the gene for ADPKD may not develop cysts that can be seen on ultrasound or CT scan for quite some time. Although the majority of ADPKD patients have detectable cysts by the time they are adults, it is only after the age of 30 years that a negative ultrasound almost certainly means that someone does not have ADPKD. Since there are 2 genes that can cause ADPKD, and since families with the 2nd gene usually have milder disease, it is conceivable that they develop detectable cysts even later in life.

Our children have been screened for ADPKD. One of our children has a cyst on one kidney. Does this mean that she has ADPKD? We have not told her about the results yet. What do you suggest?

While solitary or simple cysts are common in healthy adults over age 50 years, a single kidney cyst in a child without PKD is very rare. For your child who comes from a family where one parent has ADPKD, the finding of a single kidney cyst most likely indicates that she, too, has ADPKD. However, sometimes other kidney structures can be mistaken for a cyst on ultrasound. Therefore it is prudent to repeat the ultrasound a few years later before confirming that your child has ADPKD. Informing children that they have ADPKD is a personal decision. Every family, every parent and every child is different with regard to receiving news about a diagnosis. Furthermore, a new diagnosis of ADPKD has a wide range of impact on a family's health and happiness. All of these things need to be taken into consideration. One area of support would be to discuss your concerns with your physician and obtain information about what the diagnosis in your child means. This information can help you to decide if and when you tell your child. It may be helpful to know that in our collective experience, most children with a chronic condition do best when they have as many

facts as possible, presented at a level that they can understand, and when they can take some responsibility for their own care. But it is important for your child to know that they can lead a completely normal life.

My child was diagnosed with ADPKD at the age of 6. I wasn't diagnosed with PKD until the age of 35. Does this mean my child is going to do worse than me and enter renal failure at an early age?

No. Surprisingly, ADPKD does not behave consistently in a bad or good way in families. Originally, we thought that there would be consistency within ADPKD families with regard to the course of their renal disease. We now know that just because a grandparent, parent, aunt or uncle does poorly, it does not mean that a child with ADPKD is going to do poorly. In fact, the opposite can be true. If everyone in the family has had an uneventful course with ADPKD, your child may have a more complicated course.

Why this variability exists is not clear, but it is one of the areas that scientists working with the ADPKD gene are trying to understand. One factor that does help predict how someone with ADPKD will fare is knowing what prompted the original diagnosis. For example, an at-risk person who was diagnosed with ADPKD based on a screening kidney ultrasound will generally have a milder course than someone diagnosed because of blood in the urine, severe flank pain or because high blood pressure was detected.

Taking Care

We know that one of our children has ADPKD. What kinds of complications occur in children with ADPKD?

1. *High blood pressure.* Perhaps the most common complication is high blood pressure. It is important for your child to have his/her blood pressure checked at least every year at the doctor's office. Adults and children rarely have symptoms related to high blood pressure, so there are often no clues that blood pressure is elevated. Occasionally, patients develop headaches related to hypertension, but this is not a usual case.
2. *Kidney infection.* One of the more serious medical problems for children with ADPKD is the development of a kidney or cyst infection. When this happens, children usually have a fever, sometimes chills, as well as pain over their back or sides. Often children develop nausea, lose their appetites and can begin vomiting. In addition, urination may change and become painful, frequent, or cause burning. These are serious warning signs and your doctor should be contacted as soon as possible.
3. *Blood in urine.* At the first sign of blood in the urine, you should contact your doctor immediately. There are steps that can minimize the amount of blood and the time the blood is present in the urine. Bed rest is the mainstay of treatment. The less active your child is, the quicker the blood will disappear from the urine. In addition, plenty of liquid intake should be encouraged. The more liquids your child drinks during this time, the less concentrated the blood will be and the faster it will be cleared out of the urinary system.

4. *Mitral valve prolapse (MVP)*. Mitral valve prolapse is a condition where one of four valves in the heart flip-flops during the heart cycle. For reasons that are unclear, MVP occurs in approximately 25% of adults with ADPKD. In children it is less frequent. In one study from the University of Colorado, about 8% of children with ADPKD had mitral valve prolapse. Generally, MVP is not a serious condition, but it can cause symptoms such as pounding of the chest, a racing heart (palpitations), and sometimes odd chest pains. These almost always are self-limiting and go away on their own. However, should these symptoms develop in your child, it is wise to consult with a doctor. There is no need to screen for MVP.
5. *Hernias*. Children with ADPKD are at increased risk for developing hernias. These out-pouchings usually occur in the groin area or around the belly button (umbilicus). Hernias usually don't cause any problems. However, there is a risk that loops of bowel can get trapped in a hernia. For this reason, most physicians recommend surgical repair of hernias as an elective procedure.

Our child was just diagnosed with ADPKD and has high blood pressure. How can we be sure that our child is getting appropriate medical care?

You can request consultation from a pediatric nephrologist who can discuss with your pediatrician how to proceed with your child's care. A pediatric nephrologist is a specialist who has been trained in the care of children with kidney diseases. Very often it is necessary to travel to see the pediatric nephrologist for an initial evaluation and then at intervals. Between those visits the pediatric nephrologist and your pediatrician can coordinate your child's specific care.

How often should our child be seen?

Your pediatric nephrologists can help tailor the frequency of visits to your child's needs. If the blood pressure needs a lot of attention, the child may need to be seen often. If your child's kidney function begins to decrease, more frequent visits may be needed.

We know that children get all sorts of illnesses. But since our child has ADPKD, how do we decide when to handle an illness ourselves and when we should call the doctor?

Here are a few guidelines to help you decide when to call the doctor.

1. Be sure to call the doctor if your child develops any illness that causes vomiting or prevents him or her from drinking adequate amounts of fluid.
2. Have your child evaluated quickly if you see dark, foul-smelling cloudy urine; if there is a fever that is not explained by a cold or some other kind of infection; or if there is diarrhea that lasts for more than two days.
3. If your child has frequent headaches, make sure that his or her blood pressure is checked.
4. Check with your child's physician before using aspirin or any of the drugs that are "Non-steroidal anti-inflammatory agents" such as ibuprofen (trade names Advil, Nuprin, Naprosyn, Motrin, etc.). These reduce the blood flow to the kidney and can be dangerous to children with kidney diseases.
5. Don't use medicines that raise blood pressure. Cough syrups and cold medicines that contain decongestants are among those drugs. Your physician or pharmacist can help you choose safe over-the-counter medicines.

Two of our four children have ADPKD. Are there important dietary precautions that we should know about that also won't take a lot of extra time to prepare?

The general rules of good nutrition definitely apply to ADPKD children. In addition, there are some dietary considerations that in the long run may protect your child's kidney function if she or he has ADPKD. Good hydration is an important first step. Never let your child become dehydrated.

Dietary salt intake can cause unnecessary increases in blood pressure. The problem with salt is that most people like its taste and most foods have salt added to them. Salt is a preservative, so any canned food, pre-prepared food, bottled sodas or food from fast-food chains are loaded with salt. Fresh fruits, salads and most fresh vegetables are healthy alternatives and their preparation is not that time-consuming. Pasta dishes and white meats such as chicken or fish can also be prepared quickly.

Coupled with the concern about dietary salt is the issue of dietary protein intake. There is lots of evidence that too much dietary protein makes the kidneys work too hard in their effort to dispose of waste products. Particular sources of protein such as red meat make the kidney work the hardest. It is important to balance these considerations with the protein needs of a growing child. We know that certain protein-rich foods like fish, chicken, turkey, egg whites, and pasta provide a good source of dietary protein and are easier on your kidneys than steak or roast beef. This does not mean that your child cannot have any red meat, but it is probably wise to reduce the amount to smaller and less frequent portions.

My child has ADPKD and often complains of stomachaches. Is this something to worry about?

This is a difficult question. We do know that children with ADPKD have stomach or abdominal pain more often than their unaffected brothers and sisters. Why this happens is not clear. However, it may be related to the ADPKD kidneys slowly changing size and shape. In children, stomachaches can also be due to a number of things that don't have anything to do with ADPKD, such as infection, indigestion, emotional upset or school phobias. It is therefore difficult to know if every ache or pain is due to ADPKD. A good rule to guide parents is, if the aches or pains don't go away in a few days or if they continue to worsen, you should consult with your doctor.

One of the conditions that we usually worry about in ADPKD is kidney infection. Usually with a kidney infection, children develop fever, lose their appetite, and complain of constant pain in the stomach, back or side. Sometimes urination is painful or associated with blood in the urine. If this combination of symptoms occurs, it is extremely important to contact your pediatrician immediately. Kidney infections in ADPKD patients need to be treated early and with the proper antibiotics.

My son has ADPKD. He loves to play soccer. Should we limit how much he plays or should he play at all? Are there any activities that should be avoided in children with PKD?

No, there is no need to limit how much he plays soccer. However, there are two important things to consider. First, he should always have access to plenty of liquids during his games and practices. It is important that people with ADPKD avoid dehydration. Second, it is important to minimize the risk of kidney trauma in ADPKD patients. We know that kidney trauma can cause blood in the urine, and bloody urine is associated with worse renal outcome. If your son is playing soccer at a highly competitive level, you might consider having him fitted with padding for his kidney area. Usually we recommend that children with ADPKD avoid strenuous contact sports such as competitive football, rugby, ice hockey and basketball. We also recommend that these children stop a particular activity if it is repeatedly accompanied by bloody urine.

My brother who had ADPKD died of a ruptured intracranial aneurysm. My son also has ADPKD. He is 12 years old. Should he be screened for an aneurysm?

Intracranial aneurysms are one of the most serious complications of PKD. Aneurysms rupture in 50% of the cases, and following an aneurysm rupture, there is only a 50% chance that person will survive. Fortunately, aneurysms are not very common in ADPKD patients, occurring in only 5%-10%. But, this frequency is at least two times higher than in the general population, where aneurysms occur in 1% -2.5%.

The aneurysms in ADPKD appear to cluster in families. So, if one family member has had an aneurysm, the risk of someone else in the family developing an aneurysm increases to approximately 20%. In ADPKD, aneurysms are usually discovered in adulthood between the ages of 20 to 40 years. Intracranial aneurysms in ADPKD children are extremely rare. Recent evidence has shown that ADPKD patients who are screened for an aneurysm and do not have one, have not developed aneurysms later.

However, ADPKD patients who have had aneurysms detected previously can go on to develop a new aneurysm. This means that aneurysms probably develop with time in ADPKD patients, and a normal image of the brain in someone who has had an aneurysm does not necessarily mean that an aneurysm won't develop at a later date. Therefore, when your son reaches his 20s, it probably would be worthwhile for him to consider being screened for an intracranial aneurysm.

My daughter, who is 16 years old, has ADPKD. She has received information about birth control and the topic of birth control pills has come up. My understanding is that birth control pill use increases her risk of developing liver cysts. What do you suggest?

Yes, there is information to suggest an association between birth control pill use and the development of liver cysts and the severity of liver cystic disease. Even so, the majority of ADPKD women get liver cysts, regardless of whether they have used birth control pills. Unlike the kidney, cysts in the liver don't harm liver function. It is also important to note that pregnancy also increases the risk of developing liver cysts. Contraceptive use in teenagers is a complicated issue, which is probably best handled by parents and teens in consultation with their physician.

Family Issues

Our children do not know that their father has ADPKD. However, his kidney function is no longer normal and he has less energy than he used to. He will probably have to start dialysis in the next year. How are we going to deal with explaining this to our children and what it means for them?

This is a difficult issue for every family with ADPKD. While you don't want your children to be upset by their father's illness, it is also something that is impossible to hide. In addition, their father's diagnosis may involve the children directly, due to the genetic nature of ADPKD. Every child and every family is different when it comes to handling news of an illness. There is no single approach that will be right for every family and every child.

One possible approach is to clarify for yourself and your spouse (possibly making a list of questions) the concerns you have regarding: 1) your children finding out their father is sick; 2) your children finding out that their father has ADPKD; and 3) your children discovering that they are at risk for the same disease. Once these issues are clarified for your family, it would then be worthwhile to discuss these concerns with your physician prior to discussing ADPKD with your children. One possibility is that either your children's doctor or their father's doctor could meet with the entire family to explain what ADPKD is all about.

It may be comforting to know that most children do best when they have as many facts as possible and when these facts are presented at a level that they can understand. From both the father's and the children's perspectives, it is important to know how much hope there is for adults with ADPKD who are heading for dialysis. In general, it is not uncommon for people to feel better after beginning dialysis, and, in particular, ADPKD patients tend to do much better than other patients on dialysis. Kidney transplantation is the best option for ADPKD patients and provides the chance of gaining back normal kidney function without the need for dialysis.

Our child has ADPKD. We have heard that children with chronic diseases like ADPKD can have problems with their family and friends. What kind of problems might we face?

If your child has normal blood pressure, normal kidney function and no problems with pain, there should be no problems related to the child's ADPKD. If the child has significant problems with high blood pressure, kidney infections, pain and/or decreasing kidney function, frequent absences from school and frequent trips to the doctor may result. Fortunately such a severe course is rare in children. Sometimes the child's brothers and sisters become upset because the parents must spend so much time with the sick child. Parents may have marital difficulties that are created by worry about the child, the financial drain of health care costs, and, perhaps, their own ADPKD-associated guilt, knowing they have passed on the gene that causes the disease. All of these factors can create stress for the family with ADPKD.

How can families get help with these problems?

Pediatric nephrology teams have experience in helping families deal with these problems. Each pediatric nephrology team includes specially trained people, such as social workers, psychologists, psychiatrists, dieticians, and school-teachers, who can help address a family's particular problems. Pediatricians who have known a family for a long time may also be able to help with some of these problems or make referrals to someone else who can help. Many families experience some of these problems. Don't be afraid to ask for help.

Being a member of the PKD Foundation and participating in its Chapters and educational programs may also be helpful. It is often comforting to network with others who face the same situation.

Research

How can my family help?

The studies of the past fifteen years have set the stage for dramatic advances in our understanding of PKD in general and ADPKD in particular. Funding agencies, philanthropists and the general public must be told how important it is to provide money for this kind of research:

There are several ways to help:

- 1) Let senators and representatives know that it is important for the government to increase funding for PKD research through the National Institutes of Health (NIH). This is vital to obtain the kind of money needed to accomplish additional breakthroughs in research.
- 2) Contact the owners and officers of businesses and corporations that donate research funds, and tell them about the work of the PKD Foundation.
- 3) Tell friends and family about the work the PKD Foundation is doing and that donations help, no matter how big or small.
- 4) Become a member of the PKD Foundation. Membership provides a direct opportunity to support research as well as obtain up-to-date information about the latest research breakthroughs.
- 5) Volunteer your time and work with other families in your community to:
 - Foster public awareness of PKD and disseminate information about PKD to patients, the public, the medical community and the media.
 - Assist patients and family members by providing encouragement, information and a listening ear.
 - Attract individuals, corporations and foundations interested in financially supporting PKD research, education, awareness and membership.
 - Lobby for increased federal funding of PKD research through the National Institutes of Health.
 - Make a difference in your community.

We have entered a very exciting phase of clinical and scientific discovery. Current research efforts are providing a clearer understanding of the basic disease processes in ADPKD, and new evidence suggests that a cure may be possible for future generations of children with ADPKD.