

Screening and Testing for Autosomal Dominant Polycystic Kidney Disease (ADPKD)

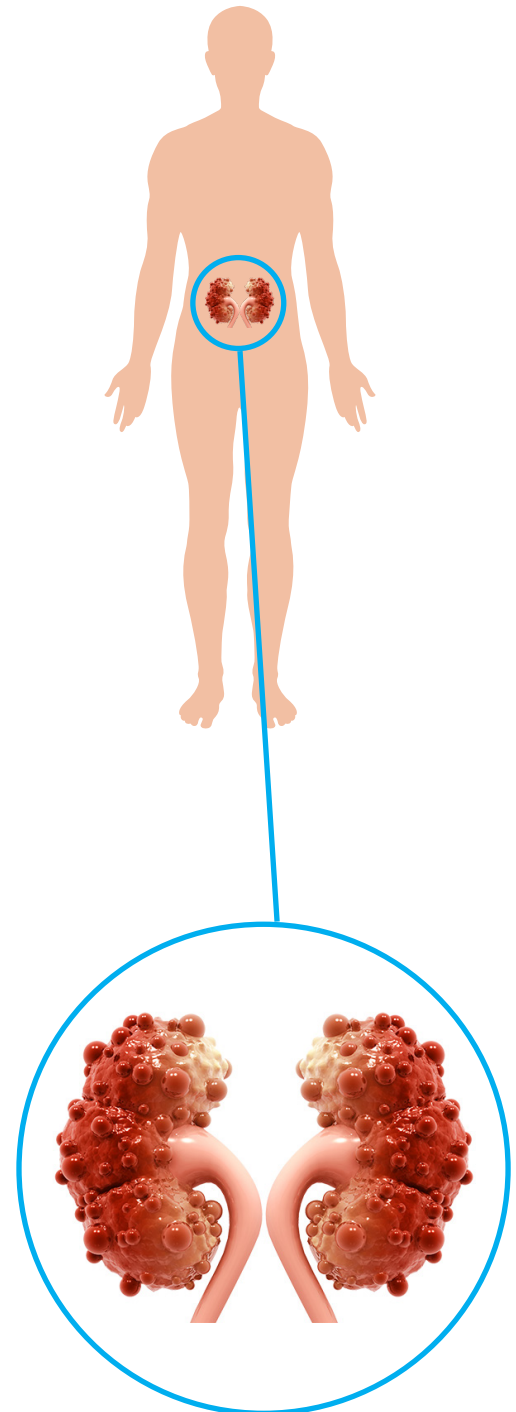
ADPKD is a genetic disorder that can affect whole families, meaning it is passed on from parents to children. In some cases, the disease may also occur randomly in a person without a family history of the disease.

ADPKD is most often caused by changes in specific genes linked to ADPKD with the two most common being PKD1 and PKD2¹, and less often by changes in other genes. Many people with ADPKD don't know which gene causes their disease. The information in this resource applies to all cases, regardless of which gene is causing ADPKD for you and/or your family members.

Not everyone who has ADPKD shows obvious symptoms. But because the condition can be inherited and affect family members, it is important to discuss family screening and testing with your kidney care team. This resource can be used to inform your decision on whether or not to pursue testing for ADPKD.

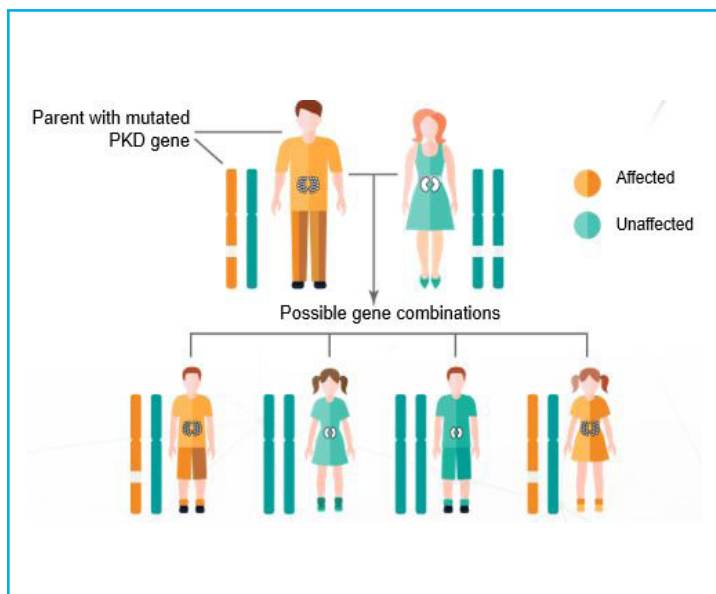
How is ADPKD inherited?

A common question among parents is whether their child(ren) will develop ADPKD. Each child who has a parent with ADPKD has a 50% chance of also developing the condition. If that child inherits the condition, they in turn will have a 50% chance of passing it on to any children they have.



Notably, it's not possible for ADPKD to "skip a generation." If your child does not have ADPKD, they cannot pass it on to their children. This means that your child must have ADPKD in order for *their children* to have a 50% chance of developing the disease.

If you have a confirmed diagnosis of ADPKD, you may want to speak to a genetic counsellor before you have children or have more children. A genetic counsellor can provide you with information to support decision-making and family planning.

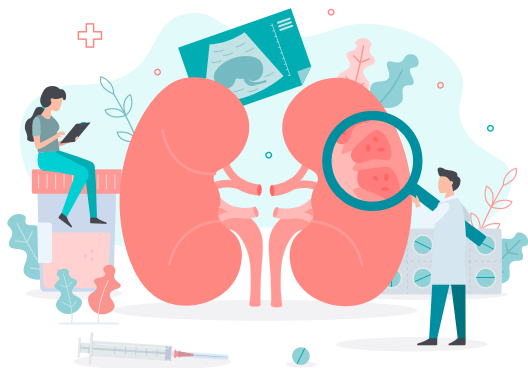


What is family screening and testing?

If one person in a family has ADPKD, this means other family members may also have the condition; looking for family members who may also be affected is referred to as screening. In many cases people with ADPKD do not have any symptoms, so the only way for them to discover they are at risk is when other family members are tested and confirmed to have it.

The best way to start screening is by talking to your immediate family members (for example parents, siblings and children), who can discuss testing with their physicians. If immediate family members are found to have the disease, screening may also be needed for extended family members (for example, cousins, aunts and uncles) but your physician can help recommend who should be tested.

How is testing done?



The most common test is an ultrasound of the kidneys. This is a simple, non-invasive test to look at the size and shape of the kidneys where features of ADPKD can be identified. It is a safe, and easy test to perform; it is very similar to the ultrasounds performed for example on pregnant women. A general practitioner (family doctor) can order an ultrasound test.

Another option is genetic testing. Genetic tests can be done to determine if a person has one of these altered (mutated) genes that leads to ADPKD, and which mutation is present.

Although ADPKD is a genetic disease, not everyone with ADPKD requires genetic testing as part of their kidney care. In fact, the majority do not get this testing done as it often does not result in any changes to their care. A genetic test may be particularly helpful if a diagnosis is unclear after imaging tests are done. Your kidney specialist can help you decide whether or not to pursue genetic testing.

If genetic testing is being done, this involves collecting a sample containing cells (and therefore DNA). This is most often done in the form of a blood test. Genetic testing can also be performed on an unborn child; in this case cells from the placenta or amniotic fluid can be sampled and analyzed.

In British Columbia, there are specific criteria used to decide if genetic testing is recommended and covered by MSP. However, the availability and cost of genetic tests may vary in other provinces. For more information on how to get tested, you can visit the PKD Foundation website at [EndPKD.ca](https://www.endpkd.ca).



Additional Considerations for Testing Younger Children and Adolescents

There are some unique points to consider in terms of screening children:

- In children, teens and young adults, a negative ultrasound test cannot completely rule out the possibility of ADPKD. In other words, the test is not 100% conclusive because cysts may still be present, but too small to detect through an ultrasound test early in life.
- In many cases, a positive ultrasound or genetic test in a *young* child is unlikely to change any management.

A common approach to screening of children is described below, but please note this is a general approach and you should discuss with your kidney care team how this applies to you and your family.

- Once children are about 5 years old or older, they may start having their blood pressure checked as part of their routine checkups. It is extremely uncommon to have high blood



Your kidney care team will determine the best approach to screening based on each individual family situation. Some other screening options that may be offered include routine physical examinations, blood pressure monitoring and genetic testing depending on individual factors.

pressure from ADPKD at this young age, but monitoring blood pressure is important for ensuring that it is in normal range.

- If their blood pressure is high or if they have symptoms like significant side or back pains, at that point they should be tested.
- If their blood pressure remains normal and they remain asymptomatic, testing can be put off until the late teens or early 20s, when they can decide as young adults whether to be tested or not.

Why should I or my family members get tested for ADPKD?

Not everyone with ADPKD or from a family known to carry ADPKD gets tested. There are some considerations to think about to help you decide whether you choose to move ahead with testing or not.

Some positive aspects to getting screened and tested include^{1,2}:

- **Feelings of relief.** Some people find relief – even if their test confirms ADPKD – at finding an answer.
- **Better lifestyle choices.** Especially for children or young adults, knowing they have ADPKD can in turn lead to more timely intervention and care. Some people who are diagnosed with ADPKD later in life may feel angry that they weren't aware of their risk for the disease earlier in life. For example, awareness of risk may encourage some young people to adopt a kidney-friendly diet, avoid medications that can damage the kidneys (e.g., nonsteroidal anti-inflammatory drugs), and start earlier monitoring and treatment of blood pressure. These measures have the potential to delay disease progression and the onset of kidney failure.
- **Earlier management of ADPKD:** There are some disease management options available to slow the progression of ADPKD, which can be most effective when started early during the course of the disease. As well, treatments to minimize complications associated with ADPKD (for example aneurysm) may be administered sooner.



- **Family planning.** Confirming whether or not you have ADPKD or the genetic mutation for ADPKD may influence your decisions related to family planning. For example, some people may choose not to have children, or choose to pursue special medical procedures to reduce the likelihood of their child developing ADPKD (see section about family planning further below).
- **Broader testing of other family members.** If your test is positive, this may prompt other family members to be tested, leading to better information and care to support them if they have ADPKD.
- **Living donation.** In families with ADPKD, sometimes members who don't have the disease decide to donate a kidney to a loved one who does have it. Screening family members for ADPKD could help identify those with healthy kidneys who may want to be donors.

There are also some challenges that may impact you and your family if you decide to get tested. These may include²:

- **Waiting for test results.** This may cause anxiety or other difficult emotions.
- **Inconclusive results.** With both imaging and genetic testing, the results can sometimes be inconclusive. This means that tests may need to be repeated, or additional tests need to be done. For some people, they may wait for several years with uncertain results.
- **Anger, shock or anxiety at the results.** If you don't have any symptoms of ADPKD and a test shows you have ADPKD or inherited the ADPKD gene, this may lead to a range of emotions. For some people, it may affect their mood and outlook on life, if they know they may live with ADPKD in the future.
- **Getting insurance coverage.** Concerns about not being able to obtain insurance, whether it be life insurance, travel insurance, etc. may make some people hesitant to get tested. There are many benefits of knowing you have ADPKD early and are being monitored by a kidney specialist. Discuss options for insurance coverage with an insurance broker for an individualized assessment.

While there are both pros and cons involved with ADPKD screening, many health care professionals will advise that, with treatments available and the advantages of early management, the benefits of screening outweigh the challenges. This is especially true as advancements in management of ADPKD continue, yielding a better quality of life for patients who receive care. For more information to support your decision, you can have a discussion with your doctor or kidney care team.

What if I test positive for ADPKD?

For people who were not aware of their ADPKD diagnosis or their genetic susceptibility to ADPKD, confirmation of the disease can cause a wide range of emotions. There is no right or wrong way to respond to this news.

Some things that may be helpful during this time include:

- Talking to a trusted friend or family member about your results
- Learning more about ADPKD from your kidney care team or a genetic counsellor
- Finding support from the [PKD Foundation of Canada](#) and similar organizations
- Knowing that, with your tests results, you will now be able to monitor your health more closely and receive more timely care, as needed.

ADPKD is a disease that affects each person very differently, ranging in severity and progression. To learn more, you can visit our webpages that offer general information about ADPKD at [BCRenal.ca](#) ► [Health Info](#) ► [Kidney Care](#) ► [Polycystic Kidney Disease](#).

Everyone with ADPKD should be seen by a nephrologist (a doctor who specializes in kidney health). If you do not have a nephrologist yet, you can ask your doctor for a referral. Speaking with a nephrologist and other members of the kidney care team during this time may help you better understand ADPKD and how to discuss this new information with your family members. If a test confirms that you have ADPKD, your family members may also want to seek support.

Family Planning

Learning that you or a family member has ADPKD may affect your decision to have children. You can learn more about this option by visiting our Pregnancy and Family Planning resource found at: [BCRenal.ca](#) ► [Health Info](#) ► [Kidney Care](#) ► [Polycystic Kidney Disease](#) ► [Managing my ADPKD](#).



Along with your kidney care team, a genetic counsellor may be able to provide you with information to support your decisions related to family planning.

References

1. PKD Charity. Genetic counselling and genetic testing in ADPKD. Retrieved December 10, 2020, from <https://www.pkdcharity.org.uk/about-adpkd/just-diagnosed/genetic-counselling-and-genetic-testing-in-adpkd>
2. A Patient Perspective on Genetic Testing for ADPKD The Lack of Complete Genetic Information, Especially Early in the Course of the Disease, Is Harming Adult Autosomal Dominant Polycystic Kidney Disease (ADPKD) Patients. <https://cjasn.asnjournals.org/content/early/2020/08/14/CJN.14051119>

This resource was developed in collaboration with the Kidney Foundation, BC and Yukon Branch, and the PKD Foundation of Canada.

