

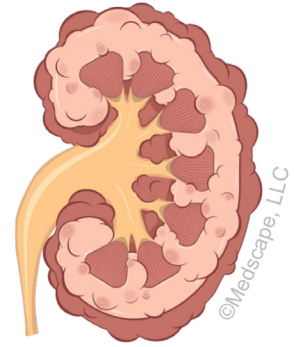
# Your Guide to ADPKD and Genetic Testing

## What Is ADPKD?

ADPKD stands for autosomal dominant polycystic kidney disease, which is a health condition where groups of cysts -- round sacs filled with fluid -- grow inside your kidneys. The cysts grow slowly over time, can be different sizes, and may grow to be very large.

ADPKD is a chronic condition, meaning people have it their whole lives. As the cysts slowly grow larger over time, they may affect how your kidneys work and may eventually cause them to stop working (kidney failure). Cysts can also sometimes grow in other areas of your body, including your liver, spleen, pancreas, colon, and the blood vessels in your brain or heart.

ADPKD is also a genetic condition. Your genes are the instructions inside your cells that determine your traits and characteristics. When there is a change, or mutation, in your genes, it can sometimes cause a condition such as ADPKD. Autosomal dominant means that if 1 parent has the condition, then that parent can pass the condition to their children.



Kidney with cysts growing throughout it

## ADPKD Symptoms

People who are living with ADPKD can have many different symptoms. More common ones can include bladder or kidney infections, kidney stones, blood in your urine, or pain in your side or back. Cysts growing outside the kidneys can cause symptoms like feeling full in the belly, heart problems, and headaches, among others. High blood pressure is also common in people who are living with ADPKD. So if you have a family history of ADPKD, you should get your blood pressure checked regularly. If blood pressure is not controlled, it can cause other problems in the body.

Although many different symptoms are possible, some people may not experience any. And different people can start experiencing symptoms at different points in their lives. However, even if you do not have any symptoms, ADPKD can still damage your kidneys over time.

## Should You Get a Genetic Test?

Your doctor may recommend a genetic test for ADPKD for a variety of reasons. If you are having ADPKD symptoms, your doctor may recommend a genetic test if you don't know whether or not you have a family history of ADPKD. If you are not having symptoms but know you have a family history of ADPKD, then your doctor may recommend a genetic test to prepare yourself or to prepare for family planning. Your doctor may also recommend that your family members get genetic tests for similar reasons.

## What to Expect With Genetic Testing



Genetic testing often involves first seeing a genetic counselor who will go over any family history you may have and can explain the genetic testing process. They can provide advice for choosing a genetic test that may be right for you and help with the insurance process. Many genetic counselors are also accessible through telehealth.

Genetic tests for ADPKD look at the genes that are associated with ADPKD, but it is important to know that your results may not be as simple as ‘positive or negative.’ There are many different genes associated with ADPKD, and each gene can

have multiple variations (called variants). Some variants are known to cause ADPKD, and some variants are known to not cause ADPKD. There are also some variants that have an unknown effect on the condition. It is best to ask your geneticist (genetics doctor) or genetics counselor for clarification no matter what your results are, but especially if your results are uncertain.

## Questions to Ask Your Doctor

- What are the symptoms of ADPKD?
- Should I have a genetic test for ADPKD?
- What do my test results mean?
- If my test results are positive, what are my next steps?
- Should any of my family members also be tested?
- Will I need to see other doctors or healthcare team members?