# How is ADPKD diagnosed

This article is a general guide for people who have been referred for tests to see if they have ADPKD.

**What different tests can be used to diagnose ADPKD?**

The way in which ADPKD is diagnosed depends, in part, on your personal circumstances. The diagnosis can be made in most people using a scan to look at their kidneys; less commonly a blood test is required for genetic testing.

If you have a family history of ADPKD, you're more likely to have the disease than other people.

- You're likely to be diagnosed using an ultrasound scan of your kidneys. We explain this type of scan later on this page.
- Sometimes, a more detailed scan of your kidneys, such as a CT scan or MRI scan, may be performed. This might be recommended if you have troublesome symptoms, for example. These scans use x-rays (CT scan) or magnets (MRI scan) to make an image of your kidneys.
- Occasionally genetic testing might be recommended too; however, not all people need this and it's something you can discuss with your doctor.

About 10 to 15 out of every 100 people (15%) with ADPKD don't have a family history of the disease. The process for diagnosis can be different if you don't have a family history of ADPKD.

- Your doctor may initially suspect that you could have ADPKD based on a scan you've had (ultrasound, CT or MRI). You may have had this scan to investigate symptoms such as back pain, high blood pressure, recurrent urinary infections or blood in the urine (haematuria).
- Alternatively, a doctor may notice your condition when you're having imaging for a different reason altogether (such as pregnancy). In this case, the diagnosis of ADPKD is often unexpected.

Although the presence of cysts in the kidneys suggests a diagnosis of ADPKD, there can be other causes of kidney cysts too. Therefore, your doctor will assess you for these other possibilities before making a diagnosis of ADPKD. Your doctor may suggest genetic testing to help confirm or rule out a diagnosis of ADPKD for you.

### Ultrasound scans

An ultrasound scan uses high frequency sound waves (which humans cannot hear) to make an image of the inside of the body (Figure 1). Using ultrasound, a specialist can examine your kidneys from outside your body.

**Figure 1: Ultrasound picture of a kidney with multiple cysts**

Several cysts are identified in this kidney. The larger cysts are marked by arrows, and appear as dark circles. In a person with a family history of ADPKD, a picture like this establishes that the person has ADPKD.

In ADPKD, cysts develop over time, and so might not be present in children or young adults. Therefore, if you're under 30, it may not be possible to use ultrasound to confirm a diagnosis of ADPKD, even if you have a family history of the condition. You may be advised to have ultrasound scans periodically repeated in the future, to check for cysts as you get older. Similarly, ruling out a diagnosis of ADPKD in people under 40 years is difficult. However, by age 40, a diagnosis of ADPKD can be ruled out in individuals who don't have multiple kidney cysts.

### Genetic tests

ADPKD is caused by a faulty (mutated) gene. Mutations in one of two genes can cause the disease: *PKD1* or *PKD2*.

Genetic testing is usually performed on sample of blood. It checks the DNA in this blood sample for faults in the *PKD* genes. Genetic testing is the most accurate way of making a diagnosis of ADPKD. It's available through the NHS, but it's not required to make a diagnosis of ADPKD in most people.

To find out more about genetic testing – and whether it might be appropriate for you – see our web page 'Genetic counselling and genetic testing in ADPKD'

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