

I want to know more about...

ADPKD in Children*

* Autosomal Dominant Polycystic Kidney Disease



pkdcharity
Polycystic Kidney Disease

together the answer

This leaflet is intended as a general guide for parents or carers of children who are at risk of, or have been diagnosed with, autosomal dominant polycystic kidney disease (ADPKD). Find out here how ADPKD can affect children as well as how it is diagnosed, monitored and treated.

What is ADPKD?

Autosomal dominant polycystic kidney disease (ADPKD) is a relatively common inherited condition in which fluid-filled cysts develop in both kidneys. The kidneys are normally filled with many thousands of thin, fine tubes called tubules that filter the blood and produce urine. In people with ADPKD, these tubules eventually become too big and fill up with fluid (like small balloons), forming cysts. The cysts press

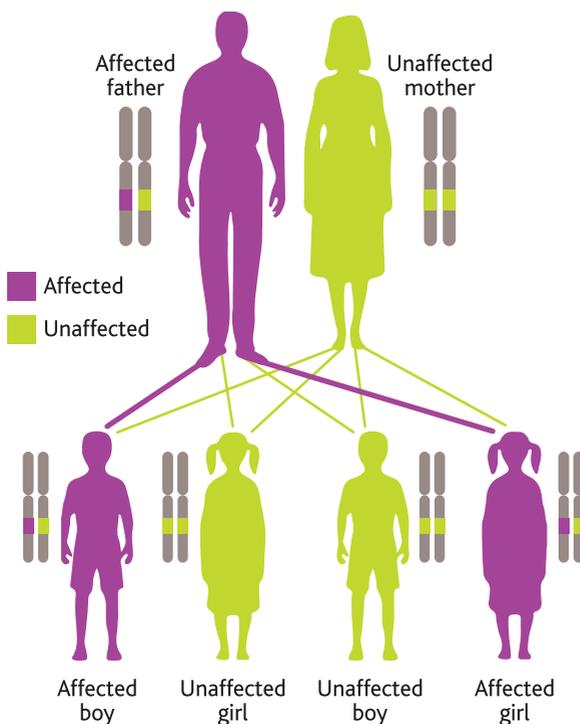
on the rest of the kidney and stop the kidney from working properly.

ADPKD is believed to affect one or two in every 1000 people. Although ADPKD causes progressive kidney failure, it very rarely causes major symptoms during childhood. If your child has ADPKD, they probably won't require treatment for kidney problems until later in life.

Is my child at increased risk of getting ADPKD?

ADPKD is caused by a faulty gene, which is usually inherited from a parent. If you have ADPKD, there is a one in two (50%) chance your child will inherit the disease. Similarly, there is a one in two (50%) chance that they will have normal kidneys. The risk is the same for every child you have.

Sometimes, a child can have ADPKD even though neither parent does. This happens when a new gene fault develops in a child. However, this is much rarer: only about six to eight out of every 100



In ADPKD, a faulty PKD gene can be inherited from a mother or father (depends on who has a faulty gene).

children with ADPKD don't have a parent with the condition.

What are the symptoms of ADPKD in children?

Most children with ADPKD don't have kidney symptoms that cause problems. The cysts are only just beginning and there is plenty of normal kidney tissue to filter their blood.

Up to a quarter of children do have some problems though. These are mostly similar to the symptoms that adults with ADPKD get.

Symptoms can include:

- ❖ Pain in the sides or back
- ❖ Urinary tract infections
- ❖ Kidney stones
- ❖ Passing more urine than normal (this can happen even with small cysts)
- ❖ Blood in the urine

About one in five children with ADPKD get high blood pressure, but this usually doesn't cause symptoms. It's important that your child's blood pressure is monitored and treated if it becomes high, however, as we explain later.

Children are more likely to get symptoms as their ADPKD gets worse and cysts become bigger. It's extremely rare for children with ADPKD to develop such severe disease that they need dialysis or a kidney transplant during childhood.

There are some signs that might suggest that your child's ADPKD will progress more quickly.

These are:

- ❖ Having enlarged kidneys
- ❖ Having high blood pressure

Your specialist will be able to explain whether your child will benefit from any additional monitoring or treatment if they have either of these findings.

How is ADPKD diagnosed?

There are two tests for ADPKD:

- ❖ Ultrasound scan
- ❖ Genetic test

In children at risk of ADPKD, the condition is usually diagnosed through an ultrasound scan of the kidneys. Ultrasound is quick, painless and safe, but doctors cannot always see cysts in their early stages. So, even if your child's scan does not find cysts, there is still a chance that they could develop cysts in later life. For this reason, ultrasound scans may be repeated at a later date.

Genetic testing uses a blood test to check for faulty *PKD* genes. If a specific *PKD* gene fault (mutation) causing ADPKD has been found in you or relatives already, it is much more straightforward to test your child for this specific mutation. (For more information on the genetics of ADPKD, see our 'Genetic counselling and genetic testing in ADPKD factsheet'.)

In most cases, an ultrasound scan or genetic test will give a definitive answer on whether or not a child has ADPKD, but neither genetics nor ultrasound are perfect and may fail to confirm the diagnosis. You should discuss this further with your doctor if you'd

like a full explanation of the pros and cons of each type of test for ADPKD.

Some parents-to-be choose to have special genetic testing before or during pregnancy. You can discuss this with your doctor if it's something that you want to consider.

Should my child be tested for ADPKD?

You may know that your child is at risk of having ADPKD because you or their other parent has the condition. Parents in this situation often ask whether they should have their child tested for ADPKD. Doctors used to believe that it was best for children to not be checked for ADPKD until they were old enough to decide for themselves whether or not they want to have the tests. Some kidney specialists and GPs may still have this view. However, experts now advise that children can be tested, provided

that their parents and carers agree with healthcare professionals that this is the best thing to do. Children should be included in this discussion when possible (depending on their age and understanding). You should be offered counselling from the NHS before deciding to go ahead with tests. Of course, after considering all of the options, it is still fine for you to choose to wait until your child is old enough to decide about testing for themselves.

The table below shows some of the pros and cons of testing for ADPKD in children. If you choose for your child not to have an ultrasound or genetic test, doctors can still check your child's health, such as their blood pressure.

Blood pressure check-ups for children with ADPKD

Experts recommend that children and young adults (up to age 18) with

Should my child be tested for ADPKD?

| Advantages of testing in childhood | Disadvantages of testing in childhood |
|--|---|
| Genetic testing usually provides an answer as to whether or not your child has ADPKD, meaning less uncertainty. | Testing during childhood will stop your child being able to decide for themselves as an adult whether or not they want to be tested for ADPKD. |
| For your child, receiving a diagnosis of ADPKD when they are younger may be easier for them to come to terms with. | Some children won't have any symptoms or signs during childhood. Knowing they have ADPKD may not make a difference to their health, although it might increase worry. |
| If more treatments become available for children with ADPKD in the future, doctors may be able to offer these to your child. | Having a diagnosis of ADPKD confirmed may have implications for your child's future life insurance and health insurance. |



Monitoring your blood pressure.

ADPKD have their blood pressure checked at least every 2 years, from the age of 5 years. This testing is also available to children with a family history of ADPKD which shows they might have a 50% risk of the disease.

About one in every five children with ADPKD has high blood pressure, and it's thought this might speed up damage to the kidney. Blood pressure checks are recommended to spot high blood pressure, so treatments can be used to get it back under control. Hopefully, this treatment can slow down kidney damage. Reducing high blood pressure is also good for general health too.

Although these checks-ups are recommended, you can choose for your child not to have them if you prefer.

Kidney check-ups for children with ADPKD

In the early stages, ADPKD is unlikely to show many symptoms, despite some changes beginning to happen in the kidneys. This can be difficult for parents because it can make it hard to tell if your child's ADPKD is getting worse. This is where regular monitoring can help.

The aim of monitoring for children with ADPKD (and those at risk of ADPKD) is to check for early signs of kidney damage so that treatments can be started early. Urine tests, blood tests and scans can check your child's kidney health and size.

In general, doctors recommend that children diagnosed with ADPKD have a check-up at least once every two years, but this will be tailored to your child's needs. This could be with your GP or a hospital doctor, depending on your local NHS services.

Children at increased risk of having ADPKD but not diagnosed can also have blood pressure checks.

If you have any questions or concerns about the frequency that your child has check-ups, discuss this with your specialist.

How can ADPKD be treated in children?

Most children with ADPKD do not need any treatment until they reach adulthood.

If your child has high blood pressure, your doctor might recommend this is treated with medication, such as an ACE inhibitor (angiotensin-converting enzyme inhibitor). This is to help reduce their risk of having heart problems or blood vessel disease in the future. It might also slow down the speed at which their ADPKD worsens, although there have only been a few studies on this. Ask your kidney specialist to explain to you the pros and cons of different medicines available for your child.



A drug called tolvaptan can help to slow down the speed of ADPKD progression in some adults, but it is very rarely prescribed to children. More research is needed before we will know whether tolvaptan is safe and helpful for children with ADPKD.

Is there anything I can do to prevent health problems in my child with ADPKD?

This question is often asked, and unfortunately doctors have few precise answers. Some helpful advice for parents of children diagnosed with ADPKD is:

- ❖ A healthy diet and lifestyle are helpful (as for all children). There's no need to radically change things if your child is diagnosed with ADPKD. Your child's kidneys will function well in the early stages of the disease, so it's fine for them to eat a normal diet.
- ❖ It's sensible to avoid your child eating too much salt because it can make blood pressure problems worse.
- ❖ If your child produces a lot of urine, they may need to drink more fluid to stay hydrated. You and they can check that they are drinking enough by looking at the colour of their urine – it should be clear to light yellow, not darker (see the colour chart to the right).
- ❖ If your child has particularly large kidneys with cysts or has a lot of kidney pain, your specialist may recommend that they avoid heavy contact sports. This is to reduce the risk of them injuring their kidneys.
- ❖ Always check with your doctor or pharmacist before giving your child any medication (even those you can buy without a prescription) to check it is safe for them. Most medications are fine, but some, such as certain non-steroidal anti-inflammatory drugs (e.g. aspirin and ibuprofen), can damage the kidneys of people with ADPKD. Ask for a clear recommendation on safe alternatives for your child.

Check your child's urine colour



Clear to light yellow

You're hydrated.
Drink as you get thirsty.



Dark yellow to dark amber

You're dehydrated.
Have a drink of water.

Strange coloured urine can be caused by some foods or medicines. It can also signal a health problem. See your doctor if your pee has changed colour and you haven't had foods or medicines to explain this.



Cloudy (milky)

You might have a urinary tract infection, especially if you have other symptoms, like a burning sensation when you pee, needing to pee more often or smelly pee. Kidney stones can also cause cloudy pee.



Pink, red or light brown

This can be caused by some medicines or food (e.g. beetroot, blackberries or rhubarb).

If you're female, it could be vaginal blood. Sometimes, a urinary tract infection causes blood in pee. Or if you have ADPKD, it could signal a cyst infection, bleeding cyst or kidney stone.



Dark brown

This can be caused by some medicines or food (e.g. broad beans, rhubarb or aloe). It might also signal a urinary tract infection or kidney or liver problem.



Green or blue

This can be caused by some medicines or food colours. It can also be a sign of a urinary tract infection or a liver problem.



Dark yellow or orange

You might be dehydrated. Orange pee can also be caused by some medicines, laxatives, B vitamins or carotene (found in carrots and some other fruit and veg) or a liver problem.

These urine colours are for illustrative purposes only and may not match the colour of your urine.

❖ Useful links and sources of other information

Genetic counselling and genetic testing in ADPKD

pkdcharity.org.uk/about-adpkd/just-diagnosed/genetic-counselling-and-genetic-testing-in-adpkd

How is ADPKD diagnosed?

pkdcharity.org.uk/about-adpkd/just-diagnosed/how-is-adpkd-diagnosed

Talking to children and young people about ADPKD

pkdcharity.org.uk/about-adpkd/living-with-adpkd/talking-to-children-and-young-people-about-adpkd

Diet and Lifestyle

pkdcharity.org.uk/about-adpkd/living-with-adpkd/diet-and-lifestyle

ADPKD and Pregnancy

pkdcharity.org.uk/about-adpkd/living-with-adpkd/adpkd-and-pregnancy

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The PKD Charity Helpline offers confidential support and information to anyone affected by PKD, including family, friends, carers, newly diagnosed or those who have lived with the condition for many years.

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For the latest version including version with references, please visit <http://www.pkdcharity.org.uk/about-adpkd/just-diagnosed/adpkd-in-children>



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