ADPKD in Children

This leaflet is intended as a general guide for parents or carers who have 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 [1,2]. 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 until later in life [1–3].

If my child is at risk of ADPKD, should they be tested?

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. Until recently, specialists usually gave the answer of 'No', because very few children required any treatment and there were not many treatment options available. It was felt safe to wait until the child was old enough to decide about testing for themselves.

Some doctors now believe that it might be helpful to diagnose ADPKD earlier. This is so children can start having treatment sooner if needed, for example for high blood pressure. Some studies show that we may be able to help to slow the progression of the disease in some patients if we know which children are affected early[2–4]. So, some doctors now recommend testing for ADPKD during childhood.

Others would still recommend a 'watch and wait' approach, where children would only be tested if they showed symptoms.

Ask your doctor to explain their recommendation, so you understand the reasoning behind it. Although specialists can give advice, you and your family will need to make the decision together.

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

Should my child be tested for ADPKD?	
Advantages of testing in childhood	**Disadvantages of testing in childhood**
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 whether or not they want to be tested for ADPKD as an adult.
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, so knowing they have the disease earlier may not help.
If more treatments become available for children with ADPKD in the future, doctors may be able to offer these to your child if their ADPKD is confirmed.	Having a diagnosis of ADPKD confirmed may have implications for your child's future life and health insurance.

It is also worth looking at 'Talking to children and young people with ADPKD' from the PKD Charity before making a decision about talking to your child about ADPKD.

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 of their parents 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 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 symptoms that cause problems. Their kidney cysts are only just beginning and there is plenty of normal kidney tissue to filter their blood [2,3,5,6].

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

Symptoms can include:

- Pain in the sides or back
- Urine infections
- Kidney stones
- Passing more urine than normal (this can happen even with small cysts) [7]
- Blood in the urine

About a quarter of children with ADPKD get high blood pressure, [1,2,8] but this usually doesn't cause symptoms. It's important that your child's blood pressure is monitored, and treated if it becomes high, as we explain later.

Children are more likely to get symptoms as their ADPKD gets worse and cysts become bigger. It's very 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 a child ***might*** have ADPKD that will progress more quickly. These are [13,14]:

- Having enlarged kidneys
- Having high blood pressure

This might not always be true though, so if your child has these symptoms, speak to your specialist to find out what it might mean for them.

How is ADPKD diagnosed?

In children at risk of ADPKD, the condition is usually diagnosed through an ultrasound scan. 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.

Genetic testing is not routinely done at the moment because it's expensive and can't detect all of the genetic faults that cause ADPKD [3]. This may change as genetic testing technology improves in the future. In the meantime, some parents choose to have special genetic testing before or during pregnancy. You can discuss this with your doctor, if it is something that you want to consider.

How can ADPKD be monitored in children?

In the early stages, ADPKD might not 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. Monitoring can check the size of your child's kidney cysts and their blood pressure. Monitoring is especially important for checking high blood pressure because it might not cause symptoms, so you may otherwise be unaware of the problem.

In general, doctors recommend that children diagnosed with ADPKD have a check-up at least once a year, 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 should also have regular checks on blood pressure and urine. Your child may also have blood tests at some visits.

Children at increased risk of having ADPKD should have the same tests.

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 [1-3].

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 more research is needed before we can be sure [9–11]. As experts are not sure how much benefit blood pressure medication is to children, ask a kidney specialist to explain to you the pros and cons of different medicines available for your child.

More research is being done to see if we can slow down the formation of kidney cysts in people with ADPKD. Although results so far are promising [16,17], more research is needed before we will know whether these medications are 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 specific answers. Some helpful advice for parents of children diagnosed with ADPKD is:

- A healthy diet and lifestyle is 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.
- 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 [2].
- 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, ibuprofen) can damage the kidneys of people with ADPKD. Ask for a clear recommendation on safe alternatives for your child.

Useful links and sources of other information

Genetic counselling and genetic testing in ADPKD

How is ADPKD diagnosed?

Talking to children and young people about ADPKD

References

1 Torres VE, Harris PC, Pirson Y. Autosomal dominant polycystic kidney disease. *Lancet* 2007;**369**:1287–301.

2 Cadnapaphornchai M. Autosomal dominant polycystic kidney disease in children. *Current Opinion in Pediatrics* Published Online First: 29 January 2015. doi:10.1097/MOP.0000000000000195

3 Sweeney WE, Avner ED. Diagnosis and management of childhood polycystic kidney disease. *Pediatric nephrology (Berlin, Germany)* 2011;**26**:675–92.

4 Grantham JJ. Rationale for early treatment of polycystic kidney disease. *Pediatric nephrology (Berlin, Germany)* Published Online First: 15 July 2014. doi:10.1007/s00467-014-2882-8

5 Mekahli D, Woolf AS, Bockenhauer D. Similar renal outcomes in children with ADPKD diagnosed by screening or presenting with symptoms. *Pediatric nephrology (Berlin, Germany)* 2010;**25**:2275–82.

6 Fick GM, Duley IT, Johnson AM, *et al.* The spectrum of autosomal dominant polycystic kidney disease in children. *Journal of the American Society of Nephrology : JASN* 1994;**4**:1654–60.

7 Helal I, Reed B, McFann K, *et al.* Glomerular hyperfiltration and renal progression in children with autosomal dominant polycystic kidney disease. *Clinical journal of the American Society of Nephrology : CJASN* 2011;**6**:2439–43.

8 Cadnapaphornchai MA. Hypertension in children with autosomal dominant polycystic kidney disease (ADPKD). *Current hypertension reviews* 2013;**9**:21–6.

9 Ecder T, Schrier RW. Cardiovascular abnormalities in autosomal-dominant polycystic kidney disease. *Nature Reviews Nephrology* 2009;**5**:221–8.

10 Cadnapaphornchai M a, McFann K, Strain JD, *et al.* Increased left ventricular mass in children with autosomal dominant polycystic kidney disease and borderline hypertension. *Kidney international* 2008;**74**:1192–6.

11 Martinez-Vea A, Bardají A, Gutierrez C, et al. Exercise blood pressure, cardiac structure, and diastolic function in young normotensive patients with polycystic kidney disease: A prehypertensive state. *American Journal of Kidney Diseases* 2004;**44**:216–23.

12 Thong KM, Ong ACM. Sudden death due to subarachnoid haemorrhage in an infant with autosomal dominant polycystic kidney disease. *Nephrology, dialysis, transplantation : official publication of the European Dialysis and Transplant Association - European Renal Association* 2014;**29 Suppl 4**:iv121–3.

13 Fick-Brosnahan GM, Tran Z V, Johnson AM, *et al.* Progression of autosomal-dominant polycystic kidney disease in children. *Kidney international* 2001;**59**:1654–62.

14 Cadnapaphornchai M a, Masoumi A, Strain JD, *et al.* Magnetic resonance imaging of kidney and cyst volume in children with ADPKD. *Clinical journal of the American Society of Nephrology : CJASN* 2011;**6**:369–76.

15 Schrier RW, Abebe KZ, Perrone RD, *et al.* Blood pressure in early autosomal dominant polycystic kidney disease. *New England Journal of Medicine* 2014;**371**:2255–66.

16 Torres VE, Chapman AB, Devuyst O, *et al.* Tolvaptan in patients with autosomal dominant polycystic kidney disease. *The New England Journal of Medicine* 2012;**367**:2407–18.

17 Cadnapaphornchai MA, George DM, McFann K, *et al.* Effect of pravastatin on total kidney volume, left ventricular mass index, and microalbuminuria in pediatric autosomal dominant polycystic kidney disease. *Clinical journal of the American Society of Nephrology : CJASN* 2014;**9**:889–96.

Published by the PKD Charity

The PKD Charity is a registered charity in England and Wales (1160970), Scotland (SC038279).
A company limited by guarantee. Registered company in England and Wales (9486245)

Information Product Ref No ADPKD.ICYP.2015V2.0

First published November 2015
Due to be medically reviewed November 2018

Authors and contributors

Written by Dr Matko Marlais, Academic Clinical Fellow in Paediatric Nephrology, Great Ormond Street Hospital or Children/UCL Institute of Child Health.
Reviewed by Professor Paul Winyard, Institute of Child Health, London.

With thanks to all those affected by ADPKD who contributed to this publication.

For further copies of this information sheet or other PKD Charity information visit www.pkdcharity.org.uk

If you don't have access to a printer and would like a printed version of this information sheet, or any other PKD Charity information, call the PKD Charity Helpline on 0300 111 1234 (weekdays, 10.00am-4.30pm) or email info@pkdcharity.org.uk

PKD Charity Helpline: 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.

Disclaimer: This information is primarily for people in the UK. We have made every effort to ensure that the information we provide is correct and up to date. However, it is not a substitute for professional medical advice or a medical examination. We do not promote or recommend any treatment. We do not accept liability for any errors or omissions. Medical information, the law and government regulations change rapidly, so always consult your GP, pharmacist or other medical professional if you have any concerns or before starting any new treatment.

This information has been produced under the terms of The Information Standard. References used to produce and review the information are available on request.

We welcome feedback on all our health information. If you would like to give feedback about this information please email info@pkdcharity.org.uk