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## **KHA-CARI Patient and Caregiver Guideline for the Diagnosis and Management of Autosomal Dominant Polycystic Kidney Disease**

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## **KHA-CARI Patient and Caregiver Guideline for the Diagnosis and Management of Autosomal Dominant Polycystic Kidney Disease**

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### **About this guideline**

The Kidney Health Australia-Caring for Australasians with Renal Impairment (KHA-CARI) Guideline group has produced this Patient and Caregiver Guideline on Autosomal Dominant Polycystic Kidney Disease (ADPKD) to provide advice and information for the detection and management of ADPKD. These guidelines are based on the current KHA-CARI Clinical Practice Guidelines which were published in November 2015 and can be found in the [KHA-CARI website](#):

The ADPKD guideline covers 12 topics which are grouped into three parts.

#### **PART I – Detection, Genetic Counselling and Genetic Testing**

1. Imaging Tests to Detect Disease
2. Genetic Testing
3. Genetic Counselling

#### **PART II – Management of ADPKD**

4. Imaging Tests for Monitoring Disease
5. Diet and Lifestyle Management
6. Drug Therapy
7. Psychosocial Care

#### **Part III**

8. End-Stage Kidney Disease
9. Kidney Stones
10. Chronic Pain
11. Intra-Cranial Aneurysms
12. Polycystic Liver Disease

The guideline is available on the [KHA-CARI website](#).



## **Development of the Guideline**

### ***Patient and Caregiver involvement***

Patients and caregivers were directly involved in the development of these guidelines. Five patients and two caregivers reviewed the proposed scope and topics developed by the guideline working group. Recommendations for additional topics were identified and included in the final scope. Draft versions of the guideline were reviewed by two patients and one caregiver. Their comments were addressed by the guideline writers.

The final draft was made available for public consultation through KHA-CARI, Kidney Health Australia (KHA) and the Polycystic Kidney Disease (PKD) Foundation of Australia websites.

Finally the guidelines were reviewed by members of the KHA-CARI Steering Committee before being finalised.

The KHA-CARI Patient and Caregiver Guideline for the Diagnosis and Management of Autosomal Dominant Polycystic Kidney Disease is located on the KHA-CARI website and disseminated through KHA and the PKD Foundation of Australia.

## **Acknowledgements**

We would like to thank the patients, caregivers and members of the KHA-CARI Steering Committee who reviewed these guidelines. We would also like to acknowledge Kidney Health Australia and the PKD Foundation of Australia for assisting with the public consultation process. Finally we would like to thank the guideline writers for their commitment to this project.



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## **KHA-CARI Patient and Caregiver Guideline for Autosomal Dominant Polycystic Kidney Disease – Imaging Tests for Diagnosis**

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### **1. How would I know if I have the condition?**

It is uncommon to have symptoms of ADPKD in people younger than 40 years of age. Symptoms that may potentially arise are pain in the back, blood in the urine, or a sense of fullness in the abdomen. You may suspect this diagnosis if you have a family member who is known to have ADPKD. To find out if you have ADPKD, you can have a scan on the abdomen. This scan is called an ultrasound, and it will basically take photos of your kidneys and other organs. This allows your doctor to look at the features of this condition, such as cysts. However, some scan results may not be helpful and you may need more tests to help confirm or exclude ADPKD.

### **2. Will the ultrasound be able to give me a clear diagnosis?**

An ultrasound can help give a diagnosis in a lot of the cases, but in some cases, especially if you are young, a clear diagnosis may not be possible. This is because the cysts in the kidney often develop slowly, and the full features of the disease may not have developed at the time of the ultrasound. There is a standard set of criteria used to make a diagnosis of ADPKD. These criteria depend on the patient's age and the number of cysts seen on the ultrasound. The test becomes less accurate for patients younger than 40 years of age. While for patients over 40 years of age, the ultrasound should be able to give a clear diagnosis.

### **3. How much will the test cost me?**

The cost of having an ultrasound will vary depending on the radiology practice. The cost is approximately \$200 to \$300 dollars. At this present time having an ultrasound will not cost you anything, as it is subsidised by Medicare. Some private radiology places have bulk billing. This means you can have it done for free. Other radiology practices may charge a fee. This amount often varies. The ultrasound is the first diagnostic test used as it is cheap and non-invasive.

### **4. What other tests can determine PKD?**

Simple tests such as regular blood pressure measurement by the family doctor, blood tests and urine tests can help detect early disease. This is of particular importance to those with a family history of PKD.

In terms of more specific tests, there are two other scans known as a CT (Computed Tomography) or a MRI (Magnetic Resonance Imaging) that may give better images of the kidneys. The MRI can detect



smaller cysts. However, these tests are expensive and involve some risks. These risks include exposure to radiation and a reaction to the contrast fluid that is injected before the scan. Sometimes these scans may still not be helpful in making a diagnosis. Finally a blood test to look at your genetic make-up (DNA) may be needed to make a clear diagnosis. This test is known as genetic testing. However it is expensive, time consuming and is not covered by Medicare.

#### **5. What is the best age to test children for PKD?**

In general terms, we do not recommend testing children for PKD, for several reasons.

- Most tests that are available (other than genetic testing) may not be able to positively make or exclude a diagnosis in children. This is mainly because the cysts may be too small to be detected.
- Currently, there is no treatment for stopping the development of the disease. Therefore, knowing the result will not change the way children are managed.
- Finally, being 'labelled' with such a diagnosis may have unwanted consequences like increased health insurance cost. So this becomes a costly exercise which will probably not do much more than create anxiety.
- Children often do not have any signs or symptoms of kidney disease. However, if there is a positive family history of ADPKD and the parent specifically wants to know, then a scan may be done or a genetic test. This should be done after careful discussion with the parents and genetic counsellor.



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## **KHA-CARI Patient and Caregiver Guideline for Autosomal Dominant Polycystic Kidney Disease – Genetic Testing**

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### **1. What is genetic testing?**

Genetic testing is a type of medical test that detects changes (mutations) in the genes that cause ADPKD. The test involves having a blood sample taken. The genetic material known as DNA from that sample is examined to find the genetic change that has caused ADPKD in the person being tested.

### **2. Is it necessary to get genetic testing done?**

Genetic testing is not indicated in every patient. It is expensive, and the results may be negative or unclear. Unclear results can happen even in families who have been diagnosed with ADPKD by their doctor. Genetic testing is suitable in the following situations:

- when a final diagnosis is needed in a young person
- to try to prove the diagnosis in a patient who has a negative family history of ADPKD
- in couples wanting genetic counselling and family planning advice
- when the disease presents in childhood or adolescence, or in very mild cases of the disease. By knowing about the genetic defect, your doctor may be able to give you an explanation or information about the development of the disease.

### **3. Is genetic testing for ADPKD reliable?**

Genetic testing for ADPKD is not perfect; however the chances of finding a change in the genes are very high when a person has a definite clinical diagnosis of ADPKD. Overall, genetic testing finds a change 90% of the time. The chance of finding it is higher if there is a family history of ADPKD than if there is no family history.

### **4. What steps should I take to get genetic testing done?**

Genetic testing is done through a Clinical Genetics unit. They also offer pre-test counselling to prepare you for the test. To see a Clinical Geneticist, you will need a referral from your GP or kidney specialist. You can find a Clinical Genetics service in your area by going to the [www.genetics.edu.au](http://www.genetics.edu.au) website.



**5. How much does genetic testing cost?**

Genetic testing is a rapidly developing area of medicine. The costs of testing have decreased a lot in recent years. Medicare does not cover genetic testing, but this may change in the future. Different laboratories charge different amounts. The cost may be hundreds or a few thousand dollars.

**6. Can I still get insurance if I have genetic testing?**

In Australia, genetic information includes your family medical history and the results of genetic tests. This information can affect your application for life cover but not for private health cover. Other life insurance products such as disability, trauma and income protection may also be affected by genetic test results. By law you must give the insurance company all health or genetic information about yourself. This might affect the insurance cover that you are applying for.





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## **KHA-CARI Patient and Caregiver Guideline for Autosomal Dominant Polycystic Kidney Disease – Genetic Counselling**

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### **1. What is genetic counselling?**

Genetic counselling is the process of providing information to individuals and families. This includes information about the genetic condition, inheritance, and genetic testing. This is to help individuals make informed medical and personal decisions about their health care. Information is given by a team of health experts with specialized training in clinical genetics and genetic counselling.

### **2. What is the risk of ADPKD for my family (siblings and children)?**

About 90% of affected individuals have an affected parent. The remaining 10% would be due to a new change in the gene. Parents and siblings of an affected individual have a 50% chance of being affected with the disease, unless it has happened as a new change. Every child of a person with ADPKD has a 50% chance of getting the disease, regardless of the gender of the parent or child.

### **3. How do I tell my siblings and children about their risk of ADPKD?**

Genetic counselling is recommended for individuals living with ADPKD. Genetic counselling provides education about how the disease is passed on. Guidance and support is also given to the person for how best to share the information with their relatives. This will increase understanding of the disease and the genetic risks for family members. It also helps to deal with many of the psychosocial and moral issues.

### **4. Should I advise my relatives to have a test for ADPKD?**

Screening for ADPKD without having clear symptoms is known as ‘predictive’ testing. This may be done with a kidney scan or genetic testing. The absence of kidney cysts by ultrasound in an at-risk person aged 40 years or older rules out a diagnosis of ADPKD. It does not rule out a diagnosis of ADPKD in an at-risk person younger than 40 years of age. It is very important to note that predictive genetic testing can be offered for at-risk persons under the age of 40 years. However, the genetic variant will need to be detected in one affected family member before their relatives, who are at-risk, can be tested.

It is recommended that at-risk family members without symptoms, who want to have predictive testing (kidney imaging/genetic testing), be referred to a Clinical Genetics service for genetic counselling. For adults (>18 years), this would assess and improve the individual’s knowledge of



ADPKD. It would help to determine the reasons for wanting to have predictive testing, and also assess the possible effects of positive and negative test results.

Advantages of predictive testing include:

- a. reduces doubt and anxiety related with being at-risk of getting the disease
- b. early diagnosis allows for early detection and treatment of complications
- c. making decisions about lifestyle, health, retirement, and employment
- d. encourages discussions on reproductive options
- e. promotes sharing information with family members
- f. helps in identifying potential unaffected related kidney donors

Disadvantages of predictive testing include:

- a. depending on the age of the person, could falsely reassure someone based on kidney imaging
- b. discrimination, especially when obtaining insurance cover (health, life, and disability)
- c. employment discrimination
- d. changes in social and family relationships
- e. psychological effect (especially anxiety) of the test and development of the disease with its future complications

Predictive testing in young people (below 18 years of age), continues to be a controversial topic with many ethical, legal, and psychosocial consequences. The general agreement is that it is not considered suitable to test children who are at risk of having the disease and who do not have symptoms. This is mainly because there are no childhood treatments or convincing medical benefits at that age. It also protects the young person's ability to agree to the test until they reach adulthood, when they can better understand the various issues of predictive testing. It is recommended that children of adults with ADPKD are referred to a Clinical Genetics service for genetic counselling when they are young adults, to discuss all the issues addressed above. There are currently no clinical indications for children at risk of ADPKD to have regular kidney scans, unless they are required to have one for medical reasons (such as investigating significant abdominal pain, high blood pressure, or blood in the urine).

## **5. If I want to, how can I plan having a family?**

### **Is IVF available to PKD patients and can it be used for embryo selection?**

The best time for identifying a genetic risk and discussing the available reproductive options is before a pregnancy. We would recommend a referral to a Clinical Genetics service for formal genetic counselling for any individual with (or at risk of) ADPKD, who is of reproductive age and wants to discuss the risks to their children and/or reproductive options. It is very important to know that



prenatal diagnosis (testing the foetus before birth) and pre-implantation genetic diagnosis (IVF + genetic testing + embryo selection) is available in Australasia. But it does require identifying the disease-causing genetic mutation in one affected family member first.

Differences in views may happen between families about the use of prenatal testing. This is mainly the case if the testing is being done for the purpose of pregnancy termination rather than early diagnosis. The decision about prenatal testing should be the choice of the parents, so discussion of these issues is appropriate, and may depend on:

- a. personal experience of the disease such as severely affected family members
- b. family history of the disease (especially if detected as a foetus or severe childhood presentations)
- c. fertility problems (such as already going through IVF)
- d. social circumstances (work, career, finances, and social/family support)

#### **6. Where can I find support for people with ADPKD?**

We suggest all patients diagnosed with ADPKD be directed to appropriate patient support groups, like the Polycystic Kidney Disease (PKD) Foundation of Australia ([www.pkdaustralia.org](http://www.pkdaustralia.org)). If suitable, they may be referred for formal genetic counselling (Genetic counselling services in Australia: [www.genetics.edu.au/Genetics-Services/genetic-counselling-services](http://www.genetics.edu.au/Genetics-Services/genetic-counselling-services)).



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## **KHA-CARI Patient and Caregiver Guideline for Autosomal Dominant Polycystic Kidney Disease – Imaging Tests for Monitoring Disease Progression**

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### **1. How often should I have imaging tests?**

We suggest that everybody has an imaging test done when they are first diagnosed with ADPKD. This test can be an ultrasound, a CT (Computed Tomography) or a MRI (Magnetic Resonance Imaging) scan. Whenever possible, MRI is the preferred option. It gives more details on the sizes of the kidneys. This can help your doctor decide on your treatment plan.

The change in the size of the kidney is an important indicator of how your disease is going. However, we do not recommend having regular scans to monitor the progression of the disease or the size of the kidneys. This is because we do not have specific treatments to prevent the cysts from growing and therefore your treatment plan is unlikely to change. A scan will only be necessary if you begin to experience new symptoms such as blood in the urine, pain or infection.

### **2. Which imaging tests should I have?**

Imaging tests available for this condition are ultrasound, CT and MRI. Ultrasound is the cheapest, and has no side effects. It is a series of soundwaves which develop an image of your internal organs. It is often the first screening test used. However it is limited in how much detail it can provide on the kidneys. This scan is also recommended for follow up, if you develop new symptoms.

The CT scan is more expensive, however the cost varies. These days, some practices offer bulk billing for CT scans. If bulk billing is not offered, you may be charged anywhere between \$150-300 depending on where you have it done. The CT scan involves radiation. Sometimes to see things better, a needle containing a colour dye may be needed. A small proportion of people may have an allergic reaction to this colour dye. The dye may also cause damage to the kidneys. This is more common in people who already have some kidney damage. CT is sometimes used in those patients whom we suspect have infected cysts, or have bleeding into the cysts. This is because it gives a better picture than an ultrasound. It is also used when the ultrasound machine is not readily available for whatever reason.

An MRI scan is the most expensive test out of the three options. MRI for assessing polycystic kidney disease is not an 'approved indication' by the Pharmaceutical Benefits Scheme (PBS). This means the entire cost will be paid by you. Most patients should expect to be charged between \$300 to \$500. It may also not be readily available and the patient will need to wait longer to have the test done.



However, it gives images with excellent resolution, and can provide detailed information about the kidney such as size, number and size of the cysts.

**3. What other test can be used to measure my disease?**

You should have your blood pressure measured regularly. It needs to be kept at the level recommended by your nephrologist. A blood test can give a rough idea of how well the kidneys are working. Urine tests measure the amount of protein such as albumin in the urine. This is a good indicator of how severe the disease is. These tests should be done every 3 to 6 months depending on how advanced the disease is.

**4. What rate will my kidney function decline? Can my age of kidney failure be predicted?**

It is difficult to determine how fast or slow your kidney function will deteriorate. Although the progression of the disease is slow, it can be quite damaging. Changes to the kidneys are also hard to measure. As mentioned earlier, this is best done with a scan and blood and urine tests. For this reason it is hard to predict at which age the kidneys will fail. However, on average people with ADPKD can start having symptoms at 30-50 years of age. Kidney failure can happen between 40-70 years of age.

**5. How regularly do I have to have my blood tested and by blood pressure taken?**

**How regularly do I have to see my doctor/specialist?**

How often the patient should be monitored depends on how advanced the disease is. At the early stages when kidney function is normal/near normal (Chronic Kidney Disease Stage 2-3a), blood pressure and a blood test should be done at the minimum of every 6 months. The patient should then be seen by the nephrologist to discuss the results. For those with clearly abnormal kidney function on blood test (Chronic kidney disease stage 3b and above), this should be done every 3 months or even more often if the disease is more advanced. If you have symptoms (loin pain, discomfort on voiding or noticing blood in urine) you must visit your doctor or kidney specialist immediately.



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## **KHA-CARI Patient and Caregiver Guideline for Autosomal Dominant Polycystic Kidney Disease – Diet and Lifestyle**

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### **1. Is there a special diet for patients with ADPKD?**

The best diet for ADPKD and for chronic kidney disease in general, is a healthy, well balanced diet. Evidence shows that a diet which has more plant-based foods, more unprocessed foods, less red meat and processed (packaged) foods is better for kidney health and long life,

The Australian Dietary Guidelines (<https://www.eatforhealth.gov.au>) can be used as a general guide to healthy eating. These include:

- a. Enjoying a wide range of nutritious foods from the five food groups;
  - Vegetables
  - Fruit
  - Breads and cereals
  - Dairy products (or calcium-fortified alternatives)
  - Meat or meat alternatives (including beans, legumes and nuts)
- b. Reduce your salt intake and limit foods containing salt. No more than 1 teaspoon of salt a day. Remember most of this salt is already in processed or packaged food.
- c. Limit your intake of foods containing saturated fat (example, the white fat in meat), added sugars and alcohol
- d. Drink plenty of water (8 glasses a day or 2 Litres is a good guide)

You do not need to cut out or limit any food group in your diet unless instructed by your doctor. They will know when this is necessary by checking your blood test results. The total amount of food that is needed differs for every individual. However the bulk of your diet should be made up of wholegrain breads and cereals, lean meat or meat alternatives, vegetables and fruit. Adequate fruit and vegetable intake is linked to better outcomes for kidney disease.

### **2. Is there an ideal weight for me?**

Maintaining a normal weight range for your age and gender is important for managing kidney disease. Losing excess body weight will help control diabetes, blood pressure, cholesterol levels and improve your overall health. Overweight patients may not be suitable for a kidney transplant. It can also be harder to get access to your veins for dialysis.

Fad diets are not recommended for maintaining a healthy body weight. They are not realistic to continue in the long run. They can do serious harm to your kidneys and to your health in general. Cutting back even a little on the foods you eat too much of can make a big difference. Often the foods



we don't eat enough of are plant-based foods, including whole grains, fruits and vegetables – which are great for filling up your plate.

### **3. Do I need to limit the amount of protein I eat?**

There is no strong evidence for limiting protein intake in people with ADPKD. The Australian Dietary Guidelines recommend that two daily serves of meat and meat alternatives is enough as the main protein source in the diet. A serve is 65g cooked lean meat, 100g fish, 2 eggs or ½ cup of legumes. Still, the average Australian intake of protein is much more than the recommended amount.

People often eat bigger portions of foods than their body actually needs – which is the case for foods like meats. If you are worried about your protein intake, it is recommended to discuss with an Accredited Practising Dietitian, who can help you develop a plan to suit you (<http://daa.asn.au/for-the-public/find-an-apd/>).

### **4. How much water should I drink?**

Although there are no specific recommendations, it is important to drink to thirst and stay hydrated. Increase your fluid intake in the following situations:

- a. When exercising
- b. In hot and humid weather, especially if it is causing sweating
- c. During and after an episode of diarrhoea
- d. If advised by your doctor

There may also be conditions where you are recommended, by your doctor or healthcare team to reduce water intake. Ask your healthcare team if you are unsure.

### **5. Should I stop drinking coffee?**

The short answer is no. Caffeine, which is most commonly found in coffee, tea and some carbonated drinks, has not been shown to effect cysts in ADPKD.

Recommendations for keeping your heart and blood vessels healthy are to have up to 200 mg of caffeine per day. This means up to 2 cups of coffee or 4 cups of tea per day.

### **6. Is exercise 'harmful' for my kidneys?**

There is no evidence that exercise of any nature will harm the kidneys. In fact, regular physical activity, improving fitness and reducing the time sitting down is very important for your health and wellbeing.

If you are involved in sports which have full physical contact (for example, martial arts, rugby etc.) routine checking of your kidneys is recommended.



Remember you do not have to pound the pavement or sweat it out at the gym to see benefits!

- a. Aim for 30-60 minutes on most days of the week. This can be broken down to 10 minute blocks throughout the day
- b. Aim to sit less
- c. Break up long periods of sitting as often as possible
- d. Do muscle strengthening activities at least 2 days per week





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## **KHA-CARI Patient and Caregiver Guideline for Polycystic Kidney Disease – Drug Therapy**

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### **1. Which medications can directly treat ADPKD?**

- a. Your doctor may prescribe medications to treat high blood pressure (such as angiotensin converting enzyme inhibitors or angiotensin receptor blockers). They may also prescribe medications to lower your cholesterol. Recent studies indicate that these commonly prescribed medications may have a small effect in reducing the rate of growth of kidney cysts in PKD. Also in more than half of the people with ADPKD the risk of developing kidney failure during life is low. These medications may be all that is needed for ‘direct treatment’.
  
- b. However, there are others with ADPKD who need more effective treatments to slow the rate of growth of the kidney cysts. At the present time, tolvaptan is the drug that has shown the most promise in research studies. An important clinical trial that was completed in 2012 showed that treatment with this drug for 3 years slowed the rate of growth of kidney cysts. This could be useful for patients with ADPKD. Although these results are promising, tolvaptan caused some side effects, and a quarter of participants taking the drug in the trial had to stop taking it. Common side effects of this drug are increased urination, thirst and dehydration. Rarely, reversible (but potentially serious) problems with liver function also occurred. For these reasons, in 2014, the United States Food and Drug Administration (FDA) recommended that more studies were necessary. These additional studies are very important to ensure the drug is safe and to prove that it is effective. The studies will be completed in Australia and the USA in 2017. In the meantime and in contrast to the FDA decision, some other countries (Europe, Japan, Korea, Canada) did not wait for these additional studies to be completed and approved the use of tolvaptan for ADPKD. However, due to the doubt with safety and effectiveness, the use of tolvaptan in clinical practice in these countries remains unclear. Thus, there are two important points to make about tolvaptan:
  - more information and proof is needed before it can be used in ADPKD;
  - as mentioned earlier, not all patients with PKD will need treatment with tolvaptan, and further studies will help determine who will benefit the most from the treatment.
  
- c. Research into new drugs to treat PKD is being actively pursued. Scientists have a much better understanding of how kidney cysts grow, and so the number of research studies being done has increased rapidly over the last 10 years. Many drugs and treatments are currently in development



and being (or have been) tested in animal models of PKD. These investigations take years of hard work to complete, and are inspected in detail before they reach clinical trials in humans. Some of the drugs have had encouraging results in animals, but for various reasons, when tested in humans, they have not been effective. In view of the amount of research that is being done, it is only a matter of time before more effective treatments for those who need it become available.

## **2. Are there other medications that can help manage or improve the outcomes of ADPKD?**

- a. Treatment of abnormal or high cholesterol blood levels with “statin” medication has been shown to decrease the risk of heart problems in patients with kidney disease, including ADPKD. Some examples of these commonly used statins are: atorvastatin, pravastatin, simvastatin and rosuvastatin.
- b. Medication for treating high blood pressure is very important in someone who has ADPKD. These drugs improve outcomes from kidney and heart problems. The preferred class of drugs to treat high blood pressure are Angiotensin Converting Enzyme inhibitors (ACEI) or Angiotensin Receptor Blockers (ARB) medications. The chemical/generic names of commonly used ACEIs are: ramipril, perindopril, quinapril, enalapril and captopril. Commonly used ARB medications are: candesartan, irbesartan, olmesartan and losartan. Your doctor will recommend the best medication to use to treat your blood pressure, and sometimes more than one medication will be required.
- c. When starting a new medication, it is important to discuss it with your doctor first. All medications have potential side effects. Their use may need to be adapted to your personal situation.

## **3. How can I help ensure that there may be future treatments for ADPKD?**

- a. Research is an important way to find out about new treatments. It can also be used to find out new ways to use current treatments. By being involved in research, you can help increase our understanding of ADPKD and how it should best be treated. To find out about research and if you can be involved, you should ask your doctor first, particularly your kidney specialist or nephrologist. They will be well informed about the current research and if you are suitable to take part. You can also keep in touch with the kidney community by becoming a member of PKD Australia (<http://pkdaustralia.org>) and Kidney Health Australia (<http://kidney.org.au/>).

## **4. What medications should be avoided for patients with ADPKD?**

- a. In general, there are no medications which have been identified to specifically worsen the symptoms or the progression of ADPKD. However, there are some medications which are



generally avoided in patients with abnormal kidney function. These medications include Non-Steroidal Anti-Inflammatory (NSAID) drugs (e.g. Ketorolac, Aspirin), anti-inflammatory drugs (e.g. Nurofen/Ibuprofen), and some other pain relieving medications (e.g. Codeine). These medications should also be avoided by patients with ADPKD. Talk to your pharmacist, doctor or health professional about any medications you are thinking of purchasing.

- b. Always tell your doctors and healthcare providers, such as your chemist, that you have ADPKD and possible kidney damage. This is important, especially when they are considering prescribing you new medication. They will need to give you information about the new drug. As well as, advice that is relevant to your needs.



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## **KHA-CARI Patient and Caregiver Guideline for Autosomal Dominant Polycystic Kidney Disease – Psychosocial Care**

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### **1. How can I cope with kidney pain?**

The type and source of pain can vary in people with ADPKD. In about 9 out of 10 people the symptoms are mild, but in 1 out of 10 people, the pain can be chronic and at times troublesome. When this happens, treating and preventing the pain can be challenging but there are many treatment options which your family doctor and kidney specialist can advise. These treatment options include using medications and non-drug treatments (e.g. psychological therapy, behaviour therapy, physical therapy). Your doctor can advise which medications are safe for you to take. Also, discuss with your doctors about psychological and practical methods to manage the physical, emotional and lifestyle impact of pain.

### **2. How can I cope with anxiety or depression?**

It is common for people with a long term medical condition to get anxious or depressed about their illness. A chronic condition can affect your emotions as it can cause changes in your daily life activities, work and relationships. We suggest you discuss how you are feeling with someone you trust, such as your family doctor or your kidney specialist. Your doctor may refer you to a specialist such as a psychologist. He/she may be able to offer psychological therapy or teach you ways to cope with anxiety or depression.

### **3. What can I do about financial issues?**

People with a chronic condition can experience financial hardship as there will be more expenses involved. Even though most of the treatments will usually be subsidised by the government, some additional expenses (such as dispensing fee for medication purchase; going to hospital and missing out on work) are often involved. If you are experiencing financial difficulty it's important to discuss this with your family doctor or kidney specialist. Your doctor can suggest some solutions and involve a social worker in your care. They may be able to help you get financial support.

### **4. How do I live with tiredness and loss of sex drive?**

People with ADPKD can feel tired and drained. This may be due to different reasons such as anaemia, lack of adequate nutrition, and side effects of treatment. On the other hand, tiredness and loss of sex drive may be completely unrelated to having ADPKD, so it is important to seek advice from your doctor about the possible reasons.



#### **5. Is there support for family members?**

Families should also have an understanding of the condition and treatment options. Patients may consider bringing family members to their doctor's appointments. This will give your family an opportunity to ask questions about any concerns they may have. You can also ask your doctor for resources, support programs, and education for families. Kidney Health Australia (<http://kidney.org.au/search?query=PKD>) is a consumer organisation where you will find some very useful resources for people living with chronic kidney disease.

#### **6. How can I cope with my concerns?**

The concerns of people with ADPKD need to be acknowledged. Talking with your doctor will promote their awareness and may encourage them to better understand and address your concerns. Patient support groups or resources may provide some strategies to cope with concerns, though these may need to be discussed with your doctor. The Polycystic Kidney Disease (PKD) Foundation of Australia (<http://pkdaustralia.org/>) is a patient organisation where you can find information and education about PKD. It also provides a forum for communicating with other people with PKD as well as advocating specific issues that are important for people with ADPKD and ARPKD, and their families.



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## **KHA-CARI Patient and Caregiver Guideline for Autosomal Dominant Polycystic Kidney Disease – End-Stage Kidney Disease**

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### **1. What is the best type of dialysis for people with ADPKD - haemodialysis or peritoneal dialysis?**

Dialysis is used to replace the function of a person's kidneys. There are two types of dialysis that can be done, haemodialysis and peritoneal dialysis. Haemodialysis removes blood from the patient's circulation through an artificial access in the arm. The blood passes through a machine which cleans it and returns it back to the patient's circulation. Peritoneal dialysis cleans a patient's blood by intermittently filling and draining the abdomen with a specially designed sterile fluid. The fluid is passed into the patient's abdomen through a catheter. Both types of dialysis can be used to treat patients with end-stage kidney disease.

The type of dialysis you have will depend on a number of things.

- Your preference
- How it will affect your daily life for example ability to work and travel etc.
- Other medical conditions such as history of abdominal surgery, or a very large kidney may not allow you to have peritoneal dialysis.
- Poor blood vessel circulation may stop you from having haemodialysis.

The decision should be made after careful discussion with your family, your doctors and other health professionals involved in your care.

### **2. When would I need dialysis?**

Most people start dialysis when their kidney function falls below 10 mL/min. This level is shown in your blood tests and is known as the eGFR. Feeling very unwell is also a sign that your kidney function is very low. Some of the signs and symptoms you may experience include weight loss, swollen ankles and legs, high blood pressure, fatigue, muscle weakness, severe nausea and vomiting, chest pain, heart palpitations etc. The reasons for when to start dialysis for patients with ADPKD are the same as for other causes of kidney diseases.

### **3. How do I know if I am a suitable candidate for a kidney transplant?**

To receive a kidney transplant, people with ADPKD must be assessed in the same manner as patients with kidney disease due to other causes. This involves checking for any heart problems, diabetes, cancer, hepatitis and HIV infection. Any person who wants to donate their kidney must have blood and imaging



tests done, to see if they are suitable. If the donor is a blood relative of the patient, it is important to rule out ADPKD. This is done by doing genetic testing and imaging tests.

**4. What happens if I want a kidney transplant? My doctor has told me that I will have to have my polycystic kidney(s) removed to create room for a transplant. Is this necessary?**

People with ADPKD have larger-than-normal kidneys which can take up a lot of room. There needs to be enough space in the abdomen to fit the new transplanted kidney. Some people will have one of their own kidneys removed before or at the time of the transplant to make sure there is enough space for the new kidney.

**5. When is the best time to have a transplant?**

The best time to have a transplant is when you need to start dialysis. This can best be done when there is a living donor kidney available. The living donor can donate their kidney directly to their family member or through a paired kidney exchange programme. You can find out more about the paired kidney exchange programme by going to this link <http://www.donatelife.gov.au/about-us/kidney-exchange-programme>. If none of these options is possible, a transplant can only take place when a suitable deceased donor kidney is available.

**6. Should I consider getting a kidney internationally?**

Getting a kidney transplant overseas can lead to financial, ethical and legal problems. The Declaration of Istanbul (<http://www.declarationofistanbul.org/>) outlines the principles and guidelines for transplantation and organ donation around the world. The document points out that organ trafficking is illegal. Organising a transplant overseas can have serious consequences. Before making any plans to have a transplant overseas, speak with your doctor, family and health professionals involved in your care.

**7. What cost will there be to the donor?**

There are some costs to the living donors. These may include time off work to recover and potential medical risks. Living organ donors can receive repayments for their time taken off work. You can visit The Department of Health link below,<sup>1</sup> to find out more about this financial assistance. Health professionals involved in assessing the donor will explain all these matters.

<sup>1</sup><http://www.health.gov.au/internet/main/publishing.nsf/Content/Leave-for-living-organ-donors>.



**8. My 30 year old brother wants to give me a kidney. How do I know if he is going to get polycystic kidney disease?**

Most people (95%) with ADPKD have inherited the disease from one of their parents. The other 5% can develop the defect while still in the womb. Siblings of the patient with ADPKD have a 50% chance of also getting the disease. Both imaging and genetic testing will need to be used to see if the brother has ADPKD.





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## **KHA-CARI Patient and Caregiver Guideline for Autosomal Dominant Polycystic Kidney Disease – Kidney Stones**

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### **1. How do they diagnose kidney stones in someone with ADPKD?**

- a. If you are worried that you may have kidney stones, we recommend that you discuss this with your doctor. A CT scan without contrast may be needed, though in some cases an ultrasound may be a better option.
- b. For patients with ADPKD, special attention should be given to identifying other causes of the symptoms which may be linked with kidney stones. Similar symptoms can occur with a urinary tract infection, a kidney cyst infection or bleeding.
- c. Symptoms of kidney stones may include:
  - Severe pain in the sides of the body, back and below the ribs
  - Pain on passing urine
  - Urine that may be pink, red or brown in colour
  - Urine may be cloudy and foul-smelling
  - Nausea and vomiting

### **2. What medications are available to treat kidney stones in people with ADPKD?**

- a. There can be changes in certain chemical or electrolyte levels in your urine which may put you at risk of developing kidney stones. These chemicals include potassium, sodium, calcium, citrate etc. and should be screened for with a blood and urine test.
- b. If changes in these chemicals are found, then specific treatment may be needed. This may prevent or slow the formation of kidney stones and help manage the symptoms. However, medications may only be effective in certain conditions. There may be some other prevention or treatment approaches which do not involve medications, such as changes to the amount of water you drink. You will need to discuss your most suitable prevention strategy and treatment with your kidney specialist. They may also refer you to an urologist, who specialises in disorders of the urinary system.



**3. I require surgery for kidney stones, is this commonly used in patients with ADPKD?**

- a. In general, the surgical techniques used for treating kidney stones are the same for people with ADPKD as for those without it. If surgery is needed, your urological surgeon can advise which options are best for you.



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## **KHA-CARI Patient and Caregiver Guideline for Autosomal Dominant Polycystic Kidney Disease – Management of Chronic Pain**

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### **1. Sometimes I have a lot of back pain. What can be done for this?**

A common feature of polycystic kidney disease is back pain. This is because the weight of the cysts can drag on the spine. Some other causes of back pain include kidney stones or cyst infection. Magnetic Resonance Imaging (MRI) is a test that allows us to see the cysts in the kidneys. It is used to prove that this is the reason for the back pain.

It is important to understand that this pain may be life-long and patients have a role in trying to manage it too. We recommend:

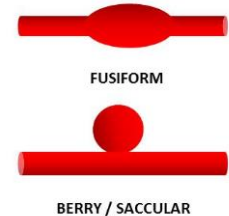
- In the first instance, try ways that avoid medications. These include backrubs, using non-steroidal creams, gels or ointments, physiotherapy, the Alexander technique, acupuncture etc. Weight loss (if you are overweight) and regular walking may also help.
- If these treatments are not useful you may need to use medication such as Paracetamol. Please be aware that you **SHOULD NOT** take more than 6 tablets of Panadol a day. That is 6 x 500 milligram tablets in total.
- There is the risk in the longer term of using stronger medicine that may be addictive. This includes medication such as Codeine, which should not be taken every day. You will need to discuss the use of stronger medication with your specialist since these require a prescription.
- If the pain is especially bad and medication does not help, then you may need to have the cysts in your kidneys removed. You should have a discussion with your specialist, surgeon and family about having surgery as there are some risks involved.



## KHA-CARI Patient and Caregiver Guideline for Autosomal Dominant Polycystic Kidney Disease – Intra-Cranial Aneurysms

### 1. What is a brain aneurysm?

This is a bulging, weak area in the wall of a blood vessel (artery) inside the brain. The bulge can be shaped like a spindle (fusiform) or a berry aneurysm (also known as a “saccular aneurysm”). This will depend on where along the artery the weakness is found.



### 2. Why do people with ADPKD get brain aneurysms?

The precise reason is unknown, but it is known that the genetic defect that causes cyst formation in the kidney is also present in the walls of blood vessels in the brain.

### 3. How likely am I to have a brain aneurysm if I have ADPKD?

People with ADPKD have a higher chance of getting a brain aneurysm (around 10%) compared to those without ADPKD (around 3%). People with ADPKD who have a family history of brain aneurysms have an even higher chance (around 15%) of having a brain aneurysm.

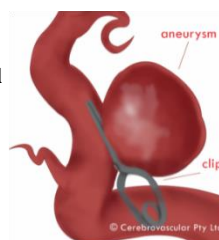
### 4. What is the best test to detect a brain aneurysm?

Most people can be tested using either a CT scan or an MRI scan. The choice of test depends on many factors such as level of kidney function and whether the scanner is available. You should discuss this with your treating doctor.

### 5. What should I do if my scan shows I have a brain aneurysm?

Your doctor can organise a referral to a specialised neurosurgical unit. They have the expertise in treating all types of brain aneurysms. How the aneurysm is managed will depend on the type and the size. Treatment options include careful observation, surgery (clipping of the aneurysm) and endovascular treatment (coiling). The specialist will explain this to you, so you can make an informed decision about the treatment.

**Microsurgical  
clipping**



**Endovascular  
coiling**





**6. How often do I need to be checked for brain aneurysms if I have ADPKD?**

Most brain aneurysms grow slowly, so testing every 5-10 years is usually appropriate. However, testing should be done anytime there are signs and symptoms of an aneurysm rupturing. For example, a sudden and extremely severe headache, blurred or double vision, nausea and vomiting, sensitivity to light, stiff neck; or stroke-like symptoms, such as weakness or numbness of the face or one side of the body, loss of vision, loss of speech, difficulty talking or understanding what others are saying.

**Acknowledgements:**

We would like to acknowledge the *Melbourne Institute of Neurovascular Diseases* for allowing us to use their images for endovascular coiling and microsurgical clipping of an aneurysm (<http://www.cerebrovascular.com.au/resources.html>).



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## **KHA-CARI Patient and Caregiver Guideline for Autosomal Dominant Polycystic Kidney Disease – Management of Polycystic Liver Disease**

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### **1. My doctor says I have cysts in my liver also. What does this mean for me?**

Cysts occur commonly in the liver, especially in women who have had children. This is because the hormone levels during pregnancy trigger cysts growing in the liver. Usually people are not affected by them. However sometimes they have a feeling of abdominal fullness or dragging. Polycystic liver disease can be detected with an abdominal ultrasound.

Women with polycystic liver disease should have counselling about the risks of pregnancy and about the use of hormone replacement therapy.

### **2. How do liver cysts contribute to my feeling of ill-health?**

Liver cysts rarely if ever cause significant liver damage in people with polycystic kidney disease. However liver cysts may cause liver enlargement and a feeling of abdominal fullness or dragging.

If there is too much discomfort, you should discuss treatment options with your doctor.

Some of these treatments include:

- a. Pain relief with other methods such as massage and acupuncture should be used first. Paracetamol should only be used if absolutely necessary. Talk about strategies with your kidney specialist, family doctor and pharmacist.
- b. Medications to stop the cysts from growing are still being tested. More information about this can be found in the guideline on drug therapy.
- c. Surgery to remove large cysts or decrease the size of the liver may be an option. However, there are risks involved. This should be considered after careful discussion with your specialist, surgeon and other health professionals involved in your care.

### **3. What other parts of the body can have cysts? How likely is the disease to spread to other organs?**

Polycystic kidney disease affects many different organs, but the kidneys are affected most often and most severely. People with disease in other organs are often not even aware of it. The liver is the second most commonly affected organ after the kidneys and may cause symptoms. The spleen and pancreas can be affected with cysts too, but usually do not cause symptoms. Sometimes people with polycystic kidney disease may develop a heart murmur, or an abdominal hernia which are also linked to the disease.