ADPKD Patient Route Map
Helping patients and families through their healthcare journey
Foreword

Using the ADPKD Patient Route Map
To use all of the features of this Route Map, you must open it using the Adobe Acrobat Reader, available to download here.

It’s best to view the Route Map in ‘Single Page’ mode. Use the interactive features below to find information and to move around the Route Map as you wish.

Use these arrows to move to the next or previous page.

Pop-ups > Click on the blue text to get further information on this topic as a ‘pop-up’ without moving pages.

These include:

Questions >
Checklists >

Hyperlinks Click on the blue, underlined text to move straight to another section with more information on this topic. In most cases these links are to other parts of the document. In some cases, the links are to websites. Hyperlinks inside pop-ups are white, but still underlined.

Patient quotes Click on these to hear from other people affected by ADPKD across Europe. Click again on the quote bubble to close it.

Navigation panel Use this panel to see where you are in the Route Map and to click to move to any other section.

Development of this Route Map
The Autosomal Dominant Polycystic Kidney Disease (ADPKD) Patient Route Map was developed jointly by the European ADPKD Forum (EAF), an independent, international group of expert doctors and patient advocates, and PKD International, the international ADPKD patient support group alliance.

The idea for this Route Map came from a Round Table meeting of expert doctors, patient representatives and organisations involved in caring for people with ADPKD. The resulting EAF multidisciplinary position statement on ADPKD care explains the basis for the Route Map.

People with ADPKD, and representatives of various ADPKD and kidney health patient organisations, provided input during the development of this Route Map.

All authors and reviewers are listed here.

Sponsorship
Otsuka Pharmaceutical Europe Ltd initiated and facilitated the EAF and funded its activities. The ADPKD Patient Route Map and the EAF Multidisciplinary Position Statement were funded by Otsuka Pharmaceutical Europe Ltd and Ipsen Farmaceutica BV.

No authors or reviewers received fees in respect of this project. This Route Map represents the opinions of the authors and not necessarily those of the sponsors.

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Suggested citation when citing and acknowledging this Route Map:
What is ADPKD?
Autosomal dominant polycystic kidney disease (or ‘ADPKD’) is a chronic condition that causes cysts to develop in the kidneys and sometimes elsewhere in the body. These cysts can cause various symptoms and they may cause the kidney to stop working properly. ADPKD is an inherited, genetic disease usually passed from parents to their children at birth. It is typically diagnosed in adulthood, but can also be diagnosed in children and even before birth.

ADPKD is a complex disease that can sometimes be overwhelming for patients and their families. In addition to the physical effects of ADPKD, the diagnosis can also have important emotional and psychological effects, including feelings of fear and anger.

Although ADPKD cannot be cured, patients, families and healthcare professionals can take various steps to help look after the kidneys and manage the effects of the disease.

Knowledge is power – and so learning about ADPKD can empower patients and carers to cope with the disease and to get the care and support they need.

What is the aim of the ADPKD Patient Route Map?
This Route Map has been developed by healthcare experts and patients to explain the types of care and support that patients and families should expect from a health service.

The aim is to help patients and families to:

• **manage their own health** as much as possible, with their healthcare team

• **talk about ADPKD** with their healthcare team and to participate in making decisions about their own care, when required

• **make the best use of available services** so that everyone affected by ADPKD gets the care, support and information they need, at the right time.

The Route Map can also help patient organisations and healthcare policymakers and providers to improve services for people affected by ADPKD.

What does the Route Map cover?
The Route Map explains what ADPKD is and how it affects patients and families. It explains how ADPKD is diagnosed, investigated and managed at each stage during the lifelong patient care pathway. It also provides advice on issues such as genetics, family planning, emotional wellbeing and finances.

The Route Map is based on the latest scientific knowledge about ADPKD and insights from experts and patients from across Europe.

Checklists are provided to help patients and families get the most out of consultations, and to help healthcare teams ensure that patients are always at the centre of their care pathway.

A list of patient organisations is provided – these are a vital source of information and support.
Knowledge

‘It’s a great help to better understand her illness.’
Peter (husband of a patient with ADPKD), Germany

‘With more knowledge and patient education, you can assume more control of the disease.’
Juan, Spain

‘Knowledge is so important – almost as important as medicine. The more you understand, the more likely you are to adhere to your treatment plan.’
Cathriona, Ireland
Cysts are fluid-filled sacs that grow and multiply in the kidneys and often in other parts of the body (especially the liver) in people with ADPKD.
Scientific knowledge

The Route Map draws on the ADPKD Consensus Conference Report published by the Kidney Disease – Improving Global Outcomes (KDIGO) initiative, the European ADPKD Forum (EAF) Report, the EAF Multidisciplinary Position Statement on ADPKD care, and other recent guidance (see Further reading).
Ongoing care and support

How the potential effects and complications of ADPKD are managed throughout life.

Lifetime ADPKD care pathway

- Diagnosis and assessment: How ADPKD is diagnosed and the initial assessments that are normally performed.
- Predicting the progress of ADPKD: Predicting the effects of ADPKD on the kidneys to help personalise care.
- Basic management and self-care: Steps that patients and carers can take, with their healthcare team, to protect the kidneys and reduce the risk of cardiovascular disease.
- Treatment to slow ADPKD progression: How some patients with ADPKD may benefit from specific treatment to slow disease progression.
- Follow-up care: Co-ordinated follow-up care involving various healthcare professionals.
- Pre-emptive: (if end-stage renal disease is predicted)
- End-stage renal disease: Role of dialysis and kidney transplantation if ADPKD causes the kidneys to fail.
- Kidney transplantation.
- Dialysis.
- Post-transplant care.

Ongoing care and support

How the potential effects and complications of ADPKD are managed throughout life.

General information

- Understanding ADPKD
- Principles of ADPKD care
- Information for health policymakers and providers
- Patient organisations
- Further reading
- Authors and acknowledgements
Understanding ADPKD

This section outlines how ADPKD can affect the health of patients.

ADPKD affects the kidneys and sometimes other parts of the body, as you can see here.

ADPKD affects different people in different ways, so not everyone will experience the effects shown here. It is also important to remember that much can be done to help reduce, manage and treat the effects of ADPKD. Although the disease can have a significant effect on life, it does not mean that people with the disease cannot live happy, long and productive lives.

Many patient organisation websites provide more information about the effects of ADPKD.

Kidneys
ADPKD is a type of chronic kidney disease that causes cysts to grow and multiply in the kidneys. If the cysts grow very large they can eventually cause the kidneys and the abdomen to swell. Cysts can eventually stop the kidneys working properly and sometimes this can lead to kidney failure, or end-stage renal disease. Cysts can also cause complications in the kidney.

If you have ADPKD, you can help to protect your kidneys from the progression of the disease through certain diet and lifestyle measures.

Why are the kidneys so important?

Wellbeing, personal and family life
ADPKD can interfere with normal activities, such as socialising, family life and work. This can have a significant emotional and psychological impact.
Symptoms

‘In my own case, living with ADPKD has not constrained me very much, as until now I have suffered from very few symptoms.’
Corinne, France

‘Due to the size of my kidneys, I look like I’m five months pregnant!’
Daniele, Germany
Cysts are fluid-filled sacs that grow and multiply throughout life in the kidneys of people with ADPKD. The number and size of cysts varies – some people have more cysts and larger cysts than others. How quickly they grow and multiply also differs between patients. The cysts usually start to develop even before birth, but most people do not notice symptoms until adulthood.
Chronic kidney disease

Chronic kidney disease (CKD) is a long-term condition in which the kidneys do not work normally or when they are damaged.

The kidneys can continue to work normally during the early stages of ADPKD, when there are fewer, small cysts. However, as the cysts grow and multiply they can stop the kidneys working properly, causing CKD (see illustration). The rate at which kidney disease progresses varies between people, as shown by the different dotted lines.

The severity of CKD is normally monitored by measuring how well the kidneys filter fluid, as explained in more detail in the Diagnosis and assessment section.

Eventually, ADPKD can cause end-stage renal disease (ESRD), the most severe stage of CKD also known as kidney failure.
Why are the kidneys so important?

The kidneys have many important roles in the body. They:

- filter the blood to remove waste products produced by the body – these are excreted in the urine
- adjust the balance of water, salts and acid in the body
- release renin, which helps regulate the blood pressure
- produce the hormone erythropoietin, which helps produce the red blood cells that transport oxygen around the body
- produce vitamin D, which helps keep the bones healthy.
At least eight out of 10 people with ADPKD develop cysts in their liver. About one in five people with liver cysts experience symptoms such as pain, feelings of ‘fullness’ and heartburn. Liver cysts can cause various complications.

Liver cysts are generally more common and worse in women than men, especially in women who have had more than one pregnancy and those who have used oestrogen medicines (including oral contraceptive pills).

A questionnaire called the Polycystic Liver Disease Questionnaire (PLD-Q) can help assess the effect of liver cyst symptoms on wellbeing.
Pancreas

The pancreas is an organ that produces digestive enzymes, which help to break down food in the gut. It also produces hormones, including insulin, that control the level of sugar in the blood.

Around one in 10 people develop cysts in the pancreas, but these do not usually cause any symptoms.
Seminal vesicles are glands that lie near the prostate. They are involved in the production of semen. Around four in 10 men develop cysts in the seminal vesicles, but these do not usually cause any symptoms or affect the sperm.
Brain (or ‘intracranial’) aneurysms occur in around one in 10 people with ADPKD, which is around three- to four-times more common than in the general public.

An aneurysm is a swelling, or bulge, in an artery. It is caused by a weakness in the wall of the artery. People with ADPKD may be at higher risk of aneurysms in large blood vessels such as the aorta (which receives blood from the left ventricle of the heart and which branches to supply the head, neck and arms) and the coronary arteries (which supply the heart itself with blood). However, these are considered rare.

Brain aneurysms do not usually cause any symptoms. In a small number of cases the aneurysm can rupture, which can be disabling or even fatal. Aneurysms are no more likely to rupture in patients with ADPKD than in other people. Generally, research suggests that each patient with an aneurysm that is conservatively treated has a 4 in 1000 (0.4%) risk of a rupture each year.

How are brain aneurysms managed?
Heart

ADPKD can cause problems with the valves of the heart, but these do not usually cause symptoms or need treatment.

Around a quarter of people with ADPKD have a heart valve problem called mitral valve prolapse. This occurs when one of the valves in the heart, called the mitral valve, does not work properly. This does not usually cause any symptoms or need treatment, and doctors are not recommended to test for this unless symptoms occur. Where treatment is needed, this can include lifestyle changes and, in severe cases, surgery.
Most people with ADPKD develop high blood pressure, which increases the risk of cardiovascular disease (such as heart attacks and strokes). Controlling high blood pressure is therefore very important.

High blood pressure usually does not cause symptoms. However, if the blood pressure becomes very high it can cause symptoms such as persistent headache, blurred or double vision, nosebleeds and shortness of breath. You should contact your doctor if you experience these symptoms. Your blood pressure should be checked from time to time.

Rarely, people with ADPKD may also have a higher risk of aneurysms in large blood vessels.
Abdominal hernias are more common in people with ADPKD than in the general population.

A hernia occurs when part of the body pushes through a weakness in the muscle that normally surrounds it. An abdominal hernia occurs in the abdomen or groin area. One study found that 45% of people with end-stage kidney disease caused by ADPKD had hernias in the abdomen, compared with 8% of those with other types of kidney failure and 4% of people without kidney failure. In some cases, surgery may be recommended to repair a hernia.

Diverticular disease is also more common among people with ADPKD who reach end-stage renal disease than among the general population.

Diverticular disease occurs when small bulges or pockets develop in the lining of the large intestine (bowel). It does not cause symptoms or need treatment in most patients. Eating plenty of fibre in the diet may help to prevent diverticular disease. Good sources of fibre include fresh and dried fruits and vegetables, beans and pulses, nuts, cereals and starchy foods.

Inflammation or infection of these pockets is called diverticulitis. The management of diverticulitis can involve laxatives, antibiotics and pain relief medication.
Erection problems are common in men with chronic kidney disease (especially end-stage renal disease), including that caused by ADPKD.

Women and men with ADPKD can also experience sexual problems related to body image issues and discomfort caused by kidney enlargement.
Principles of ADPKD care

This section explains some principles of good ADPKD care that patients and carers should expect.

ADPKD is a chronic disease which currently cannot be cured. To help limit its effects, patients should have access to lifelong care that involves:

1) A comprehensive assessment that takes into account the effects and complications that ADPKD can cause throughout the body, the likely course of the disease (i.e. prognosis) and the impact of the disease on the patient and the family.

2) Access to treatment (as clinically appropriate) to relieve symptoms, manage complications, preserve kidney function, lower the risk of cardiovascular disease, and maintain patients' quality of life.

3) Information and support to help patients and their families with recommended self-care measures and to deal with the impact of the condition.

ADPKD care should be patient-centred and involve a range of specialist healthcare professionals, according to each patient's individual needs. Specialist care should start as soon as possible after diagnosis.

Who is involved in the healthcare team?

All patients with ADPKD should have access to a nephrologist who understands ADPKD and the various ways it affects patients and families. This may be a nephrologist in a hospital or clinic.

In certain situations, referral to a nephrologist who specialises in ADPKD may be helpful to provide particular types of care, such as predicting the disease prognosis and helping to slow this where possible, managing complications and prescribing specific medication to slow ADPKD progression, and to provide opportunities for patients to participate in research.

Patients may also need access to various types of doctors and healthcare professionals with expertise in ADPKD. This is sometimes called 'multidisciplinary' care – the figure shows the healthcare professionals who may be involved.

How is ADPKD care organised?

Who is involved in ADPKD care?

<table>
<thead>
<tr>
<th>Diagnosis and assessment</th>
<th>Nephrology</th>
<th>ADPKD specialist</th>
<th>General and follow-up care</th>
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<tbody>
<tr>
<td><strong>Geneticist</strong></td>
<td>Nephrologist</td>
<td>Prognosis</td>
<td>Primary care physician</td>
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<tr>
<td>• Genetic testing and counselling</td>
<td>• Adult or paediatric</td>
<td>• Complications</td>
<td>(GP or family doctor)</td>
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<tr>
<td><strong>Radiologist</strong></td>
<td>Nephrologist</td>
<td>Medication to slow</td>
<td>Paediatric</td>
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<tr>
<td>• Detailed imaging (kidney, liver, etc.)</td>
<td>• Follow-up care</td>
<td>ADPKD progression</td>
<td>• Care of infants, children and young people</td>
</tr>
<tr>
<td>• Brain aneurysm</td>
<td>Nephrologist</td>
<td>Research</td>
<td>Psychologist / psychiatrist</td>
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<td><strong>Liver surgeon</strong></td>
<td>ADPKD specialist</td>
<td>Potential role in some</td>
<td>• Psychological effects (e.g. depression, anxiety)</td>
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<tr>
<td>• Severe liver complications</td>
<td>• Prognosis</td>
<td>aspects / complications</td>
<td><strong>Social services</strong></td>
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<td><strong>Transplant surgeon</strong></td>
<td>ADPKD specialist</td>
<td>and research co-ordination</td>
<td>• Impact on daily living, finances, family, etc</td>
</tr>
<tr>
<td>• Kidney transplantation</td>
<td>• Complications</td>
<td><strong>Manifestations / complications</strong></td>
<td><strong>Cardiologist</strong></td>
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<tr>
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<td>• Medication to slow</td>
<td>• Heart complications</td>
<td>• Chronic pain</td>
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<td><strong>Liver surgeon</strong></td>
<td>ADPKD specialist</td>
<td>ADPKD progression</td>
<td><strong>Hepatologist</strong></td>
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<td>• Research</td>
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<td><strong>Liver cyst complications</strong></td>
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<td><strong>Neurosurgeon</strong></td>
<td><strong>Pain team</strong></td>
<td>• Kidney stones</td>
<td><strong>Physiotherapist</strong></td>
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<tr>
<td>• Aneurysms</td>
<td>• Chronic pain</td>
<td><strong>Urologist</strong></td>
<td>• Anaesthetist / pain specialist</td>
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<td><strong>Psychologist / psychiatrist</strong></td>
<td>• Anaesthetist / pain specialist</td>
<td><strong>Obstetrics / gynaecology</strong></td>
<td><strong>Physiotherapist</strong></td>
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<tr>
<td>• Psychological effects (e.g. depression, anxiety)</td>
<td>• Physiotherapist</td>
<td>• Prenatal advice</td>
<td><strong>Psychologist / psychiatrist</strong></td>
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<tr>
<td><strong>Social services</strong></td>
<td><strong>Urologist</strong></td>
<td>• Pregnancy complications</td>
<td>• Psychological effects (e.g. depression, anxiety)</td>
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<tr>
<td>• Impact on daily living, finances, family, etc</td>
<td>• Kidney stones</td>
<td><strong>Dietician</strong></td>
<td><strong>Social services</strong></td>
</tr>
<tr>
<td>• Renal diet education</td>
<td><strong>Obstetrics / gynaecology</strong></td>
<td>• Dietary education</td>
<td>• Impact on daily living, finances, family, etc</td>
</tr>
</tbody>
</table>

PKD International

EAF European ADPKD Focus
Deciding about me

‘No decision about me, without me!’

Daniel, Spain
What does ‘patient-centred’ mean?

Patients should be placed at the centre of their care and treatment journey. Patients have crucial roles in managing their own ADPKD throughout their lives. Patients and families are given the knowledge and opportunity to act as fully informed partners in making decisions about their own care and about healthcare policies, services and research related to ADPKD.
Nephrologist

A nephrologist is a doctor who specialises in diseases of the kidney.
How is ADPKD care organised?

ADPKD care can involve various medical specialists and healthcare professionals. The organisation and co-ordination of these services varies between countries and regions.

ADPKD care is normally led by a nephrologist. In some situations, referral to a nephrologist with specialist expertise in ADPKD may be helpful. A team approach where all specialists in ADPKD care work in the same centre or clinic is considered beneficial. This is often not available in practice, although most university hospitals should be able to provide most of the services that may be needed. Where a local nephrologist or hospital cannot offer all services necessary, patients may be referred to other specialist ADPKD services.
What is the European Reference Network for Rare Kidney Diseases?

The European Reference Network for Rare Kidney Diseases (ERKNet) was launched in 2017 to promote high-quality, multidisciplinary care for rare kidney diseases, including ADPKD.

The ERKNet links together expert paediatric and adult nephrology centres in many European countries. A full list of the centres is here. These centres use uniform clinical guidelines and pathways, monitor the quality and outcomes of treatment, provide education for nephrologists, and support research. The network also offers ‘virtual consultations’ for doctors who need advice and provides links to information for patients.

There is also a European Reference Network on Rare Liver Diseases (RARE-LIVER), including polycystic liver disease.
Early and accurate diagnosis of ADPKD allows patients and doctors to take steps to manage the disease. Typically, ADPKD is diagnosed and assessed via a pathway like this.

## Identifying people with ADPKD
ADPKD is usually first suspected or identified in people who:
- have typical symptoms of ADPKD
- are examined for another reason, such as high blood pressure or pregnancy
- are tested (or 'screened') for ADPKD because someone else in their family has the disease.

ADPKD is often first suspected by general practitioners (family doctors).

## Diagnosis and kidney assessment
ADPKD is normally diagnosed by a nephrologist. Important initial assessments include scans and kidney function tests.

### Kidney scans
Kidney scans allow doctors to see and measure the cysts caused by ADPKD.

### Kidney function tests
Tests carried out on the blood and urine show to what extent ADPKD is affecting the function of the kidneys.

## Other investigations
It is important to find out if ADPKD has affected other parts of the body. Important investigations include:
- Blood pressure
- Liver
- Brain aneurysm

Patients may be referred to other types of specialist doctors and healthcare professionals, depending on their needs.

Should other family members be checked? >

[Checklist] >
Diagnosis

‘The PKD Association has helped me in many ways to re-orientate after my unexpected diagnosis. Most importantly it has helped to take away my fears about the disease.’

Stefan, Germany

‘I did not know I was a polycystic patient. I had a scan because at the age of 36 I had hepatitis A. The doctor told me that I had cysts on both kidneys. When I was diagnosed, I was not surprised. I suspected that there was a genetic disease in my family because all the aunts on my father’s side of the family had ended up on dialysis. At the time of the diagnosis, the doctor asked me how many children I had. “I have two children” I replied. He was very surprised and said: “... and you are not in dialysis yet?” I touched wood. It’s been 20 years since that day, and every day I am thankful because whatever happens I have been luckier than any expectation.’

Tina, Italy
Are there special issues for children?

ADPKD is typically identified in adults, but it may also be diagnosed in children. Infants and children with kidney cysts should be referred to a paediatric nephrologist. ADPKD can be difficult to diagnose in children using imaging alone. A genetic test is sometimes used to confirm the diagnosis if imaging results are unclear.
Symptoms of ADPKD

In adults these include pain in the abdomen, side or back, or blood in their urine. In children these include bedwetting and urinary tract infections.
A nephrologist is a doctor who specialises in diseases of the kidney.
Kidney scans

Kidney scans allow doctors to see and measure the cysts caused by ADPKD.

An ultrasound scan is usually used first. This is a simple imaging method that most clinics can do routinely.

Magnetic resonance imaging (MRI) may also be used. This is a more detailed, accurate and expensive scan performed in a hospital or clinic radiology department.
Kidney function tests

Kidney function tests include the measurement of the level of creatinine in a sample of blood. Creatinine is a substance produced naturally by the body. Doctors use the creatinine level to estimate the glomerular filtration rate (eGFR). eGFR is an important measure of how well the kidneys are doing their job in removing waste products and excess fluid from the blood. The part of the kidney that does this filtering job is the glomerulus, hence the name ‘glomerular filtration rate’. The eGFR falls as kidney function gets worse.

There are five stages of chronic kidney disease, defined by the eGFR.

<table>
<thead>
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<th>Stage</th>
<th>Description of kidney function change</th>
<th>eGFR level (ml/min/1.73m²)</th>
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<tr>
<td>1</td>
<td>Normal kidney function</td>
<td>90 or higher</td>
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<td>2</td>
<td>Mild loss</td>
<td>60–89</td>
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<tr>
<td>3a</td>
<td>Mild to moderate loss</td>
<td>45–59</td>
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<tr>
<td>3b</td>
<td>Moderate to severe loss</td>
<td>30–44</td>
</tr>
<tr>
<td>4</td>
<td>Severe loss</td>
<td>15–29</td>
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<tr>
<td>5</td>
<td>Kidney failure or end-stage renal disease</td>
<td>Less than 15</td>
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The urine is also tested for the presence of blood (called haematuria) or protein (proteinuria), both of which can indicate kidney damage.
Blood pressure

It is very important to identify and control high blood pressure, to reduce the risk of cardiovascular disease (such as heart attacks and strokes).
Liver

All patients with ADPKD should be assessed for liver cysts using imaging (usually by ultrasound at first). People with liver cyst complications should be referred to a hepatologist (a doctor who specialises in diseases of the liver), as necessary.
Should other family members be checked?

ADPKD is an inherited, genetic disease. People diagnosed with ADPKD should be offered counselling about the benefits and risks of informing other family members and offering them the opportunity to be checked for the disease. This screening can be done by a general practitioner, with referral to a nephrologist to confirm the diagnosis.

Checking for ADPKD allows family members with the disease to get the advice, treatment and support they may need, as early as possible. However, a diagnosis of ADPKD can have important lifelong effects, including psychological and financial consequences. Counselling can help in balancing these advantages and disadvantages when making decisions.

Routine screening for ADPKD is not recommended for children (under 18 years) who do not yet have any signs or symptoms of the disease, even if one or both parents has ADPKD. This is because of the negative psychological and financial consequences that the diagnosis may have. Children who do have signs and symptoms of ADPKD should of course be checked and referred to a paediatric nephrologist if they have cysts.

Pre-implantation genetic diagnosis (PGD) can be used to test for genetic mutations linked with ADPKD in embryos created by in vitro fertilisation. This is discussed more in the section on Family planning.
Brain aneurysm

Routine screening of all patients for brain aneurysms is not recommended because most aneurysms have a low risk of rupture and because surgery to prevent aneurysms from rupturing carries risks.

However, screening is recommended in patients with long life expectancy who 1) have a family history of aneurysms or bleeding in the brain, 2) have had a previous rupture, 3) are members of high-risk professions (e.g. airline pilots) and 4) are anxious about aneurysms even after receiving adequate information.
## Checklist: Diagnosis and assessment

- **Kidney scan**
  - Ultrasound performed and results explained
  - MRI performed (where available and necessary) and results explained

- **Kidney function tests**
  - Kidney function tests (blood and urine) performed and result explained
  - Chronic kidney disease stage explained

- **Other investigations and issues**
  - Blood pressure: tested and results discussed
  - Blood lipid (cholesterol) tests
  - Review and advice on diet, smoking and lifestyle
  - Liver: liver scan performed (when appropriate) and results discussed
  - Brain: possible need for screening discussed
  - Genetic testing: genetic testing discussed where relevant

- **Family screening**
  - Counselling provided on suitability and implications of screening other family members and available options for this service

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**Notes and questions you would like to ask your healthcare team**

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<th>Question 1</th>
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<th>Question 7</th>
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Basic management and self care

This section explains the steps patients can take, together with their healthcare team, to help protect their kidney function and reduce the risk of cardiovascular disease.

Lifestyle and diet

Few specific diet or lifestyle measures have yet been proven to prevent or slow the development of cysts in people with ADPKD. However, if you have ADPKD you can do many things that may help protect your kidney function and lower the risk of high blood pressure and cardiovascular disease.

Many of these are general healthy lifestyle measures also recommended for everyone else. They include:

• Drink more water to stay fully hydrated, which may protect kidney function in ADPKD.

• Stop smoking.

• Maintain a healthy body weight and do some form of regular exercise.

• Eat a healthy diet.

• Drink less caffeine (e.g. in coffee or cola drinks) and less alcohol.

You might be advised to make other lifestyle and diet changes if you reach end-stage renal disease.

Focus on high blood pressure

Controlling long-term high blood pressure (hypertension) is very important because high blood pressure increases the risk of cardiovascular disease, such as heart attack and stroke. Controlling high blood pressure may also help to slow the growth of kidney cysts in some people with ADPKD. In people with brain aneurysms, controlling high blood pressure (and stopping smoking) can reduce the risk that the aneurysm will burst.

How can blood pressure be controlled?

If you have high blood pressure, the lifestyle and diet measures above are particularly important to reduce the risk of cardiovascular disease.

Doctors can also prescribe various medicines to help control high blood pressure.

Regular blood pressure checks are important to make sure treatment is working.

Other risk factors

Your doctor may recommend you take other medicines to control other risk factors for cardiovascular disease.

What about complementary or alternative therapies?

Keeping it up!

Caring for your own health is very important. Maintaining a healthy lifestyle and diet, and taking prescribed medicines according to the instructions, can be difficult to maintain over long periods. Your healthcare team should be able to provide further sources of help and support locally and online. Family, friends and patient organisations can also provide valuable help and advice.

Wellbeing, personal and family life

Patients and families can take steps to limit and deal with the effects that ADPKD can have on wellbeing, personal and family life. If you have ADPKD, or are a parent of a child with ADPKD, you may wish to discuss any such problems with your healthcare team so that any necessary information, care and support can be provided.
Self care

‘Every time I go to my doctor, I realise I’m not alone and that there are many other people in my situation coping with ADPKD. If they are managing to overcome it, so will I.’

Claudia, Spain

‘I accepted it and said, “All right, let’s get married. I want to have children. I’m going to start a low sodium diet. I want to lose a bit of weight. And that will benefit me because it is beneficial for my kidneys.” And these plans helped me to have a sense of control in my life.’

Brenda, the Netherlands

‘Now my husband with ADPKD knows he will need to drink more and he has learned to calibrate the salt intake a bit better before competitions and he is very happy with the result. He doesn’t even notice the difference much, because he always used to drink a lot, and he doesn’t have any problems needing the toilet at night.’

Flavia, Switzerland

‘In principle, live as normally as possible! Research is likely to provide more possibilities to deal with the disease and perhaps prevent kidney failure one day.’

Alexander, Austria
Smoking

Smoking cessation helps to reduce the risk of cardiovascular disease (i.e. coronary heart disease and stroke) and cancer. Practical help and support to stop smoking may be available.
Body weight and exercise

Maintaining a healthy body weight and regularly exercising are recommended to help prevent and control high blood pressure.

Exercise: this can include walking, gardening, dancing and all kinds of sports – although it may be sensible to avoid high contact sports to avoid trauma to the kidneys.
Diet

Salt reduction
Recent research showed that higher dietary salt intake caused greater kidney growth in patients with ADPKD. The researchers studied data from ‘HALT-PKD’; a clinical trial of the effect of certain blood pressure medicines on the progression of ADPKD. They concluded that moderate salt restriction (to no more than 6 g a day) is beneficial in ADPKD, but you should not remove salt from your diet entirely. You may be referred to a dietician to provide a diet plan.

The recommended salt levels are lower for children.

<table>
<thead>
<tr>
<th>Age</th>
<th>Salt per day (sodium equivalent)</th>
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<tr>
<td>1–3 years</td>
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<td>4–6 years</td>
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<td>7–10 years</td>
<td>5 g (2 g)</td>
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<tr>
<td>11 years and over</td>
<td>5–6 g (2–2.4 g)</td>
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Moderate protein
There is no good evidence that low-protein diets slow the progression of ADPKD. Adults with ADPKD are advised to eat the same, moderate amount of protein (0.75–1.0 g per kg of body weight per day) recommended for the general population. Guidelines for general chronic kidney disease care recommend that adults eat no more than 0.8 g of protein per kg of body weight daily when their estimated glomerular filtration rate (eGFR; see kidney function tests) falls below 30 ml/min/1.73 m². People at risk of CKD progression are recommended to avoid a high protein intake (>1.3 g/kg/day). Any restriction on dietary protein should preferably involve education by a renal dietician and monitoring to avoid malnutrition.

Fibre
Eating plenty of fibre in the diet may help to prevent diverticular disease.

Sources
See Further reading. Some patient organisation websites provide further information about diet and ADPKD.
Caffeine

Keeping caffeine intake to a moderate level (2 cups of coffee or 4 cups of tea, per day) may be advisable for general cardiovascular health, although there is no evidence that it affects kidney cyst growth in ADPKD.
Medicines to control high blood pressure

Many different types of medicines (sometimes called ‘antihypertensive’ drugs) can be used to treat high blood pressure. Doctors consider various factors when choosing a blood pressure medicine for an individual, including the presence of other diseases.

Usually, medicines called angiotensin converting enzyme (ACE) inhibitors and angiotensin receptor blockers (ARBs) are the recommended first choices for people with ADPKD. ACE inhibitors have names ending in ‘pril’, such as enalapril, lisinopril, perindopril and ramipril. ARBs have names ending in ‘sartan’, such as candesartan, irbesartan, losartan and telmisartan.

If these are not appropriate, or if additional medicines are necessary, then other medicines called beta-blockers, diuretics or calcium channel blockers may be considered depending on the individual circumstances. Your doctor might prescribe a combination of medications to control your blood pressure.
Blood pressure checks

The standard blood pressure target for people with ADPKD is a reading no higher than 140/90 mmHg. However, the target should be personalised, taking into account age and other diseases. It may help if you know your own blood pressure target and to monitor it at home. You should discuss with your doctor what action you should take if your readings are higher than your target.

You can measure your own blood pressure at home using simple electronic devices. In certain situations, you might be given a special device to continuously monitor your blood pressure for a period at home.

This can help to provide further information about your blood pressure at different times of the day. It is important to learn how to take these measurements properly and to provide your doctor with the measurements at your clinic visit.
What do blood pressure readings and targets mean?

Blood pressure readings are given as two numbers, followed by the units by which blood pressure is measured, known as ‘mmHg’ (millimetres of mercury).

The higher of the two numbers (called the systolic blood pressure) is the pressure measured when the heart beats. The lower number (the diastolic blood pressure) is the pressure when the heart rests between beats.

Blood pressure targets indicate the readings that each number should not exceed. The typical target of 140/90 mmHg means that the systolic blood pressure should be no higher than 140 mmHg and the diastolic no higher than 90 mmHg.

Blood pressure targets should be personalised for individual patients, taking into account age and other diseases. For example, a lower target may benefit younger people with ADPKD (i.e. those aged 15–49 years) who have good kidney function and cardiovascular disease or diabetes (a disease that causes the level of sugar in the blood to be too high).
Other risk factors

Other medicines that may be prescribed to help control risk factors for cardiovascular disease (depending on individual circumstances) may include:

- medicines to control high cholesterol levels (such as statins)
- low-dose aspirin to help stop the blood from clotting.
What about complementary or alternative therapies?

Complementary (or ‘alternative’) therapies include various treatments and practices that are not part of mainstream or conventional medicine. There is no good evidence that any complementary or alternative therapy helps protect the kidneys or slows the progression of ADPKD. Some herbs used in traditional herbal medicines may damage the kidneys.

Certain types of complementary non-medical therapies may help you to cope with the effects of ADPKD and are sometimes used in pain relief.

You are advised to ask your doctor before using any complementary therapy and not to stop a treatment prescribed by a doctor on the advice of a complementary practitioner without discussing it with your doctor.

Usually you will be required to pay for complementary therapies yourself.
Checklist: Basic management and self-care

For details on these aspects please refer to the Route Map itself.

☐ Lifestyle advice provided on the following topics, with support as necessary
  – Staying hydrated
  – Stopping smoking
  – Maintaining a healthy body weight
  – Getting enough exercise
  – Eating a healthy diet, including lowering salt (sodium) intake
  – Reducing alcohol to recommended limits

☐ Blood pressure and other cardiovascular risk factors
  – Blood pressure tested and discussed
  – Lifestyle measures discussed and agreed
  – Choice of prescription medicine explained and agreed, where necessary
  – Target explained and agreed
  – Appropriateness and options for home monitoring discussed

☐ Other cardiovascular risk factors
  – Need for cholesterol-lowering treatment, or other therapy, discussed

☐ Wellbeing, personal and family life
  – Impact of ADPKD on personal, wellbeing and family life discussed, where necessary, and action taken

☐ Family planning issues discussed and addressed

☐ Information about patient organisations and other forms of support

Notes and questions you would like to ask your healthcare team
Predicting the progress of ADPKD

This section explains how the effects of ADPKD on the kidneys can be measured and the future progression estimated, to help personalise the care provided.

Cysts grow and multiply throughout life in people with ADPKD. The prognosis – that is, the rate at which the disease progresses and the effect it has on the kidneys – varies between patients. In some people, cysts grow and multiply so slowly that serious kidney disease (including end-stage renal disease) may only happen very late in life, or not at all. However, in other people the disease progresses more rapidly. This may be affected by ADPKD management, as well as each patient’s individual disease.

The rate that ADPKD progresses can be measured, and even estimated in advance. This can be useful to:

- identify patients with rapidly progressing disease who may be suitable for certain treatments or for clinical trials
- evaluate whether treatment is effective
- plan for kidney transplantation or dialysis, later in the disease course.

The progression of kidney function can be assessed in several ways.

**Family history**

Patients are at increased risk of rapid ADPKD progression if other members of their family with the disease reached end-stage renal disease before the age of 58 years. It is recommended that ADPKD progression should be checked every 3–5 years in diagnosed patients with this family history.

**Kidney function tests**

The most important factor in the prognosis of ADPKD is kidney function. Good kidney function suggests a better prognosis than bad kidney function regardless of cyst growth. Doctors can predict how kidney function will change based on repeated measurements of creatinine levels in the blood and urine (see kidney function tests).

However, people with ADPKD can have normal kidney function for many years, even though their cysts continue to grow and multiply. For this reason, doctors also use other tests to monitor and predict ADPKD progression. Two methods that may be used are: the total kidney volume (TKV) and the Predicting Renal Outcomes (PRO) PKD score.

**Total kidney volume**

Total kidney volume (or TKV) is a measure of the swelling of the kidneys caused by cysts. It is calculated from kidney scans and can be used to assess ADPKD progression.

**PROPKD score**

The PROPKD score is a research tool that predicts the risk of progression according to four factors. One of these factors is the genetic mutation present and the use of the PROPKD score is limited by the cost and availability of genetic testing. This score is currently used only for research and not for the routine management of patients.
Prognosis

‘I found out I had a polycystic kidney at the age of 35, I already had two children and I was told I would soon be on dialysis. As a matter of fact, I am now 68 years old, and my overall condition is fairly good.’
Antonia, Italy

‘When I was informed about my ADPKD it was a brutal shock and difficult news to accept. Luckily, soon after, I met a nephrologist who still follows me today and who managed to instil hope and to turn the gloomy picture into something more positive. It is crucial that doctors take time with their patients when the pathology is first announced in order to further bring hope and support.’
Corinne, France

‘Although kidney failure was predictable in my case, it was not clear when it would actually happen. In principle, the process from the first notable restrictions to kidney failure took about 10 years. The diagnosis was clear, there were no secrets. For years there were no treatment options other than blood pressure adjustment and diet, followed then by the “radical cure” of dialysis and transplantation. The output of the latter is certainly not predictable. In my case, the additional cyst growth in my liver – which was initially not prognosticated – was an aggravating circumstance. This was even more dramatic than kidney disease, because of its size and the associated massive complaints.’
Elisabeth, Austria
The total kidney volume (TKV) is calculated based on imaging from kidney scans.

- Ultrasound scans are inexpensive, widely available and can be used to estimate TKV in some situations.
- Magnetic resonance imaging (MRI) scan is the ideal method for measuring TKV, as this is more accurate than ultrasound. However, some patients may have limited access to MRI scans, especially for repeated TKV measurement. Faster and simpler MRI techniques are now increasingly available.

Doctors take into account the height and age of the patient when assessing TKV. This is because the size of the kidney means different things in a small person compared with a larger person, and in a young person compared with an older person.

Ideally, TKV should first be measured around the time of diagnosis and then at a later point.

TKV can be used in two ways to assess ADPKD progression:

**Risk prediction**
A single TKV measurement can be used to predict how quickly ADPKD will progress in the future. This may be used to make decisions about treatment. Most patients with ADPKD can be classified into one of five risk categories for disease progression (called 1A–1E) based on their TKV, adjusted for their height. In general, patients in Classes 1C–1E will have rapid disease progression.

**Repeated measurement**
Repeating the TKV measurement several times allows doctors and patients to see how quickly the kidney volume is actually changing and progressing. The availability of repeated TKV measurement varies so you may wish to discuss this with your nephrologist.

It is recommended that patients whose TKV increases more than 5% per year should be considered to have rapidly progressing ADPKD. This assessment should preferably be based on three or more measurements using MRI scans, each at least 6 months apart.

The prediction of ADPKD progression is normally conducted in centres that have a special expertise in ADPKD. A list of expert centres within the European Reference Network for Rare Kidney Diseases can be found here.

The length of the kidney (rather than the volume) can also be measured by ultrasound and used to predict whether ADPKD is likely to progress rapidly.
PROPKD score
The PROPKD score predicts the risk of ADPKD progression according to whether the patient:
1) is male or female
2) needed treatment for high blood pressure before the age of 35 years
3) had any of the main kidney complications of ADPKD before the age of 35 years and
4) has ADPKD caused by a genetic mutation called PKD1.
Checklist: Predicting the progress of ADPKD

☐ Progression of ADPKD explained
☐ Reasons for prognosis prediction explained
☐ Available options for prognosis prediction explained

Notes and questions you would like to ask your healthcare team

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How is ADPKD inherited?

ADPKD is usually caused by abnormalities (or mutations) in one of two genes: these are known as PKD1 and PKD2. In fact, ADPKD is the most common genetic kidney disease.

ADPKD is inherited in a dominant pattern. This means that the ADPKD mutation need only to be present in the inherited genes from one parent for it to cause ADPKD.

ADPKD is not the same as autosomal recessive polycystic kidney disease (ARPKD), which is a separate, rare disease with a different inheritance pattern.

Genetic testing

Genetic testing can identify if either of the PKD1 or PKD2 mutations are present. It is done by testing a small sample of blood or saliva.

Usually doctors do not need to use genetic testing to diagnose ADPKD, but it can help the diagnosis in some situations.

Pre-implantation and prenatal genetic testing are also possible for the purpose of family planning, although the availability of these tests varies.

Genetic testing for ADPKD is complex and expensive and is normally performed by geneticists in centres with appropriate experience. However, genetic tests are becoming quicker and less expensive, thanks to advances in technology.

Counselling

A positive genetic test for ADPKD can have lifelong consequences for patients and their families. As well as the effects of the disease itself, ADPKD can also affect other aspects of well-being, personal and family life, and finances.

Ideally, everyone who undergoes genetic testing for ADPKD (and the parents of tested children) should have access to counselling to discuss the advantages and disadvantages in detail. This counselling should be with a clinical geneticist, specialist nurse or genetic counsellor with expertise in ADPKD.

Counselling should also be available to discuss the test result and its implications. A child who is sufficiently mature and competent may be included in the counselling.

You can find more information about genetics and testing at the websites of ADPKD and genetic disease patient organisations.
Genetics

'I was diagnosed by pure chance when I was aged 23. I have a “de novo” ADPKD mutation – the first case in my family. I didn’t think much about it when I was young, but now that I’m 45, and after having two kids, I can’t stop thinking about it. My daughter is only 10 and was diagnosed when she was barely 2. I have huge feelings of guilt for having transmitted this disease. The mental stress out of it is so persistent and so strong that sometimes I think I am the disease … I am scared that my son could also be affected. I wish there was more psychological support for patients.”

Silvia, Italy

‘In my family of origin, four out of six siblings are affected. The disease was passed on to us by our father; it’s not known whether he inherited the disease or whether a new mutation occurred in his case. My daughter is also affected by the disease. She is already aware of this, since she has learned from experiencing my decline, and my dialysis and transplantation. It is my wish, however, not to make the disease a permanent topic in our house. It is important that she can live as carefree as possible regardless of the disease and, above all, that she chooses her career aspiration freely, regardless of the disease.

Miriam, Austria

‘In my family it is difficult to talk about the disease because everyone is highly anxious about the risk of transmitting it to the next generation. With my kids I decided to talk openly and in full transparency but without undue dramatisation.’

Corinne, France

‘I was diagnosed when I was 16. One day after a competition I went to the toilet and there was a lot of blood in my urine. I was so scared! They took me to hospital and did a lot of checks. In the beginning the doctor thought I had kidney cancer, then we found out it was something else. After that, my father and older brother were diagnosed with the same disease, but nobody told me that I had a genetic disease and I never understood that! It was only many years later when a doctor told me “What? You are having children. Are you crazy? Don’t you know you have a genetic disease?” I was shocked … It took me weeks to come to terms with this fact. I felt so guilty. Fortunately, my wife had a very positive outlook towards this thing. I would like to tell everyone that I am now 47, we have a very normal life and except for that very first trip to the hospital I’ve never had any issues with PKD!’

Rolf, Switzerland
Genes

Genes are sections of DNA contained inside almost all the body’s cells. They act as a set of instructions, telling the cells how to make the proteins that build and regulate the body. Most genes are inherited and so determine the characteristics that are passed from parents to their children. Mutations in the genes can cause a wide variety of inherited diseases. ADPKD is usually caused by one of two genetic mutations – these are known as PKD1 and PKD2. PKD1 mutations are most common, causing around three-quarters (75%) of cases. ADPKD caused by PKD1 mutations is generally more severe and rapidly progressing than that caused by PKD2 mutations.

Current genetic tests fail to identify a PKD1 or PKD2 mutation in around one in 10 people with ADPKD. In a few of these cases, PKD may be caused by other genes. Recent studies have shown that mutations in several other genes can cause ADPKD. The PKD in these cases is usually mild.
Dominant pattern

ADPKD is inherited in a ‘dominant’ pattern, meaning that the ADPKD mutation need only be present in the inherited genes from one parent for it to cause the disease. This means that:

- If either of your parents has ADPKD, there is a one in two (50%) chance that you will have inherited the disease.
- If you have ADPKD, there is also a one in two (50%) chance that each child you have will inherit the disease.

Children who do not inherit the abnormal gene causing ADPKD will not develop the disease or pass it to their children. ADPKD cannot ‘skip’ a generation, but sometimes people with ADPKD can go through life without being diagnosed and still pass the disease to a child.

In a small number of cases (fewer than one in 10), genetic mutations causing ADPKD occur spontaneously in patients for the first time, i.e. without there being any previous history of the disease in the family. Any children of these patients will still have a 50% chance of inheriting the disease.
Situations where genetic testing can be helpful include:

- where the diagnosis is unclear, especially in infants and children or if there is no family history of ADPKD
- to help predict the prognosis of ADPKD (e.g. using the PROPKD score)
- to make sure the abnormality is not present in a potential living kidney donor
- pre-implantation genetic diagnosis, which is used to test whether embryos have ADPKD genes prior to in vitro fertilisation.
Checklist: Genetics and genetic testing

- Inheritance of ADPKD explained
- Role of genetic testing discussed, where indicated
- Local availability of genetic testing discussed, where indicated
- Genetic counselling offered
- Information about patient organisations and other forms of support

Notes and questions you would like to ask your healthcare team
Treatment to slow ADPKD progression

This section explains how some patients with ADPKD may benefit from specific treatment to slow disease progression.

In recent years, research has been performed into medicines to slow the progression of ADPKD. Currently, one medicine is licensed for use in Europe and others are being studied in clinical trials.

Who is suitable for treatment?
The medicine that is licensed for use is called tolvaptan. It can only be used by adults with ADPKD who have normal or moderately reduced kidney function (stage 1–3 chronic kidney disease) at the start of treatment, and who have evidence of rapidly progressing disease.

This treatment is not available everywhere. Patients may wish to discuss its availability, and your suitability, with your nephrologist. Experts recommend that patients should be fully involved in deciding on whether treatment is appropriate for them.

What is the benefit?
Evidence suggests that, if the benefit shown in a clinical trial was maintained, this would mean that each 4 years of treatment would delay the occurrence of end-stage renal disease by approximately 1 year. The treatment may also reduce kidney complications and pain, and increases urine production. It does not affect liver cysts.

This treatment does not replace the need for other aspects of basic ADPKD management and self-care.

What are the main side effects?
The most common side effect of this treatment, occurring in all patients treated, is an increased need to urinate. If you take tolvaptan, you should take precautions to ensure you do not become dehydrated.

If you experience difficulty in urinating, this may indicate kidney or bladder problems and you should stop taking the treatment and immediately contact a doctor or go to the nearest hospital.

This treatment may cause the liver to stop working properly. You should tell your doctor if you have any symptoms that might indicate liver damage. Blood tests to check the liver function need to be done each month for the first 18 months of treatment and every 3 months after that.

How is treatment taken?
This treatment is taken in the form of tablets, twice daily. You should follow the prescribing doctor’s instructions carefully, and check with your healthcare team if you have any questions. Further information can be found in the patient information leaflet or online.

Who provides this type of treatment?
This treatment must be started and monitored by a doctor who is experienced in managing ADPKD and who understands the risks of therapy and the necessary monitoring.

Are any other treatments being developed for ADPKD?
Other medicines are being investigated to slow the progression of ADPKD and polycystic liver disease, and eventually these may provide more treatment options.

Some of these medicines are being tested in clinical trials. Patients who wish to participate in clinical trials should ask their nephrologist about the available opportunities.

✔️ Checklist >
Tolvaptan

Tolvaptan is a type of medicine called a vasopressin-2 antagonist. It works by blocking the action of the hormone vasopressin, which is involved in the development of kidney cysts in ADPKD.
Rapidly progressing disease

How ‘rapidly progressing disease’ is defined, and therefore which patients are eligible for treatment, can vary in different countries. European experts have published guidelines on which patients should be considered for treatment. National guidelines are also available in some countries (e.g. the United Kingdom here and here).
Precautions

If you are taking tolvaptan, to avoid dehydration during treatment you should always have access to water and be able to drink enough when you feel thirsty and to prevent thirst (e.g. by always having water to hand).
Symptoms that might indicate liver damage

You should inform your doctor immediately if you have signs that could indicate potential liver problems such as nausea, vomiting, fever, tiredness, loss of appetite, pain in the abdomen, dark urine, jaundice (yellowing of skin or eyes), itching, or joint and muscle pain with fever.
Checklist: Treatment to slow ADPKD progression

☐ Role and local availability of treatment discussed

☐ Eligibility for treatment assessed, based on progression assessment

☐ Eligible patients should be informed about how the treatment works and the potential benefits and risks – allowing patients to be fully involved in deciding whether to start treatment

☐ Patients who receive treatment should be instructed about:
  – how to take it
  – actions to take in the event of side effects, including side effects needing medical attention, who to contact in an emergency, liver effects and the need for blood tests for liver function monitoring, urination and water intake
  – Interactions with other medicines

☐ Discussion of other opportunities for participation in clinical trials

☐ Information about patient organisations and other forms of support

Notes and questions you would like to ask your healthcare team

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Managing kidney complications

This section explains the main complications of ADPKD that can affect the kidneys, and how these can be managed.

**Kidney cyst infections**

Kidney cysts can become infected by bacteria. This can cause fever and pain in the abdomen. Cyst infections can be difficult to diagnose. A type of scan called positron emission tomography (PET) is sometimes used.

Antibiotics are the main treatment for cyst infections.

In some cases cysts may be drained, either by surgery or a procedure in which a needle is passed into the kidney through the skin.

**Cyst rupture and bleeding**

Kidney cysts can sometimes burst (or ‘rupture’) and bleed, causing blood to appear in the urine. In most cases bleeding resolves without treatment within 7 days.

You may self-treat pain using over-the-counter painkillers. If necessary, doctors may prescribe other pain relief medicines. If you have severe or persistent bleeding you should seek medical advice.

**Kidney stones**

People with ADPKD are at increased risk of kidney stones. Kidney stones are caused by the build-up of certain chemicals in the urine. Larger stones can block parts of the urinary system and cause discomfort and pain. They may make you need to urinate more often than normal, or blood to appear in the urine.

You should contact your doctor if you think you have a kidney stone. Stones are best diagnosed using a computed tomography (CT) scan, where this is available. X-ray or ultrasound scans may also be used. Urine and blood tests may be performed, and stones that are passed in the urine may be examined.

Small stones may be passed in the urine, without treatment. Drinking extra water to increase the urine flow can help to flush out stones. For larger stones, medical help is necessary.

You may self-treat any pain using over-the-counter painkillers. If necessary, doctors may prescribe other pain relief medicines.
Positron emission tomography

Positron emission tomography (PET) is a type of scan that uses small amounts of radioactive materials, a special camera and a computer to provide detailed images of the organs and tissues inside the body.
A computed tomography (CT, or CAT) scan uses x-rays and a computer to make images of the organs and tissues inside the body. These images are more detailed than those from standard x-rays. A substance known as a contrast dye is sometimes given by mouth or injection to help make the images clearer.
Antibiotics

Antibiotics are medicines used to treat infections caused by bacteria. First-line antibiotics often used for cyst infections include members of the fluoroquinolone class (e.g. levofloxacin) and trimethoprim-sulfamethoxazole, although the choice depends on various factors and can differ between different countries and hospitals.

Cyst infections can be difficult to treat and they can return even after a course of antibiotics. It is important to take antibiotics according to the instructions given.
Urinary system

The urinary system includes the kidneys, the tube (ureter) that carries urine to the bladder, the bladder and the tube (urethra) that carries urine from the bladder out of the body.
Larger stones need medical care, and you may need to be referred to a urologist (a doctor who specialises in diseases of the urinary system).

Possible treatments for larger kidney stones include:

- Extracorporeal shockwave lithotripsy: this uses ultrasound to break up stones so that they can be passed in the urine.
- Ureteroscopy: a thin, flexible telescope with a laser is passed upwards through the urinary system and used to break up stones.
- Percutaneous nephrolithotomy: a thin, flexible telescope is passed into the kidney through a small cut in the skin and used to break up stones.
Managing liver and brain complications

This section explains the complications of ADPKD that can affect the liver and brain, and how these can be managed.

Liver

Liver cysts do not cause symptoms, or need treatment, in most patients. However, they can become infected and, when large, can cause significant pain and discomfort.

Did you know?
A questionnaire called the Polycystic Liver Disease Questionnaire (PLD-Q) can help assess the effect of liver cyst symptoms on wellbeing.

Patients with liver cysts causing symptoms should be referred to a hepatologist.

Cyst infections

Liver cyst infections can cause pain in the abdomen, and fever. When severe, they are best diagnosed using a scan called positron emission tomography (PET).

Cyst infections are treated mainly with antibiotics.

Reducing the cyst ‘burden’

If you have polycystic liver disease, it is important that you and your hepatologist agree on the goal of treatment, because this will guide the treatment options that are most appropriate for you.

Surgery can be used to reduce the number and size of liver cysts causing severe symptoms.

Liver transplantation is an option for some patients with very severe liver cysts, but is rarely needed.

New medicines are being investigated to help treat liver cysts. If you are interested in joining a clinical trial you should discuss this with your hepatologist.

Brain aneurysm

Patients found to have intact brain aneurysms should ideally be treated by a multidisciplinary team, including a neurosurgeon and neurovascular radiologist.

Surgery is sometimes performed to help prevent an aneurysm from bursting (rupturing) by stopping its blood supply.

A burst aneurysm causes bleeding in the brain. You should call an ambulance if you or someone you know with ADPKD has symptoms that you think might be caused by a ruptured brain aneurysm.

Other sections explain the management of effects that ADPKD can have in the kidneys and elsewhere in the body.

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Positron emission tomography (PET) is a type of scan that uses small amounts of radioactive materials, a special camera and a computer to provide detailed images of the organs and tissues inside the body.
Antibiotics are medicines used to treat infections caused by bacteria. First-line antibiotics often used for cyst infections include members of the fluoroquinolone class (e.g. levofloxacin) and trimethoprim-sulfamethoxazole, although the choice depends on various factors and can differ between different countries and hospitals.

Cyst infections can be difficult to treat and they can return even after a course of antibiotics. It is important to take antibiotics according to the instructions given.
Hepatologist
A hepatologist is a doctor who specialises in diseases of the liver.
Goal of treatment

The goals of treatment may include reducing the size of the enlarged liver, but also improving your quality of life and/or reducing the symptoms caused by the liver cysts. Topics for discussion include: how long should the treatment be given, and how long should it take for the goals to be achieved.
A neurosurgeon specialises in the diagnosis and surgical treatment of disorders of the nervous system, including the brain, spinal cord and other nerves throughout the body.
Neurovascular radiologist

An interventional neurovascular radiologist specialises in ‘minimally invasive’ image-based surgical procedures (or ‘interventions’) used in the diagnosis and treatment of diseases of the head, neck and spine.
Surgery: Liver cysts

Several procedures may be used to treat liver cysts, depending on the situation. It is recommended that these procedures should only be carried out by surgeons with specific expertise in polycystic liver disease.

**Aspiration and sclerotherapy**

Aspiration is a procedure in which cysts are drained by inserting a needle through the skin, under local anaesthetic. Sclerotherapy involves flushing the cyst with a chemical solution (usually alcohol) to help stop the cyst filling up with fluid again. Aspiration sclerotherapy is normally used to treat patients with symptoms caused by a large dominant cyst (typically around 5 cm in diameter, or larger).

**Fenestration**

After a cyst is cut open, part of its wall is removed so that it does not swell up again. This can usually be done using ‘keyhole’ (or laparoscopic) surgery, needing only a small cut in the skin. Having fenestration can prevent a liver transplant being needed in the future.

**Segmental liver resection**

Surgery can be performed to remove parts of the liver where severe cysts are localised. This is normally done only if other treatments cannot be used or do not work.
Surgery: Aneurysms

Surgery is sometimes performed to help prevent an aneurysm from bursting by stopping its blood supply. The two main methods are:

- **Clipping**: a metal clip is passed through a small opening in the skull and used to seal off the aneurysm.
- **Coiling**: a tube is inserted into an artery in the leg or groin, passed to the aneurysm and used to block it with metal coils.

These procedures carry risks and so need careful consideration. The risks depend on the individual situation. If you are diagnosed with an unruptured aneurysm you may wish to discuss the risks with your healthcare team, especially a neurosurgeon or neurovascular radiologist.

It is recommended that patients with untreated small aneurysms should be assessed again every 6 to 24 months.
Symptoms: Aneurysm

The symptoms of a ruptured aneurysm can include severe headache, sickness, blurred double vision and loss of consciousness.
Managing pain

This section explains how pain is assessed and managed in ADPKD, particularly when it is persistent.

Pain is the most common and important symptom of ADPKD for many patients, particularly when it persists. Pain can occur at any stage of the disease, even early on, and it is important to recognise, investigate and treat it.

What causes pain in ADPKD?

Acute pain can be caused by various kidney or liver complications of ADPKD, such as cyst infections and kidney stones. Treating the underlying cause of these should help to relieve the associated pain.

Chronic pain normally means pain lasting for more than 3 months. Chronic pain can result from the growth of cysts in the kidney or liver, and can be particularly difficult to treat.

Assessing pain

Doctors and other healthcare staff may not always appreciate how much pain can affect people with ADPKD. Ideally, doctors and nurses should routinely ask about pain at each clinic visit. You should make sure you report pain to your healthcare team, carefully explaining the location, symptoms and impact of any pain you experience.

Did you know?

Pain scales can be useful to help measure and monitor the severity of pain. Questionnaires have also been developed to help patients and doctors to assess the impact of ADPKD on patients’ wellbeing. You may wish to ask your doctor about these.

How should chronic pain be managed?

Different types of doctors and healthcare professionals often need to work together to treat chronic pain. Depending on the cause and type of pain, this may include pain specialist doctors, neurologists, radiologists, as well as nephrologists and hepatologists. Physiotherapy and psychotherapy may also have a role in some situations.

Experts in the Netherlands have produced a pathway for the stepwise management of chronic pain caused by ADPKD (see figure below).

Treatments are recommended to be used in the order shown below until pain relief is achieved.
Pain

‘I find that my renal consultant has a hard time understanding the chronic and acute pain I get due to ADPKD. It is often brushed aside.’
Anonymous, UK

‘I am lucky not to have encountered renal or back pain since I have been diagnosed, but I do believe that practising sport regularly helps in managing the pain.’
Corinne, France

‘In my own experience I have found that hypnotherapy can help to reduce anxiety and pain.’
Brenda, the Netherlands

‘No one should have pain in their life – more should be done to try to eradicate pain in ADPKD patients!’
Francisco, Spain
Pain scales

Common ways that patients can rate their own pain include:

- **Visual analogue scales:** you use these to indicate how severe your pain is on a line between two extremes, such as 'no pain' and 'pain as bad as it could be'.

  ![Visual Analogue Scale](image)

- **Numerical rating scales:** these are like visual analogue scales, but use numbers to rate the severity of pain, usually between 0 and 10.

  ![Numerical Rating Scale](image)
Neurologist

A neurologist is a doctor who specialises in treating diseases of the nervous system, which includes the brain, spinal cord and the nerves throughout the body.
A radiologist is a doctor who specialises in diagnosing and treating disease and injury using medical imaging techniques such as x-rays, computed tomography (CT), magnetic resonance imaging (MRI), positron emission tomography (PET) and ultrasound.
A nephrologist is a doctor who specialises in diseases of the kidney.
Hepatologist
A hepatologist is a doctor who specialises in diseases of the liver.
Physiotherapy

Physiotherapy is a physical form of treatment used to help people affected by injury, illness or disability through movement and exercise, manual therapy, education and advice.
These include physiotherapy, massage, ice and heat pads, and Alexander technique (which focuses on body posture and movement).
Acupuncture
This may help with some types of pain but has not been tested in ADPKD.
Psychological and educational

These approaches can be important, e.g. cognitive-behavioural therapy (CBT) and psychotherapy.
Pain-relief medicines

Pain-relief medicines (analgesics) are often used in a ‘ladder’, starting with milder medicines and working up to stronger ones if necessary, as follows:

- Acetaminophen (paracetamol) is often used first.
- Non-steroidal anti-inflammatory drugs (NSAIDs; e.g. ibuprofen and diclofenac) or mild opioids may be used if pain relief is insufficient. However, NSAIDs can damage the kidneys and they are not recommended for use by patients whose kidney function is impaired – you can find more information about their use in ADPKD here. If NSAIDs are suitable, they may be combined with paracetamol.
- Strong opioids (e.g. morphine) may be used when other medicines do not provide enough pain relief. Possible side effects of opioids include constipation, nausea, vomiting, sedation and mental changes. These medicines can also lead to psychological dependence (addiction). However, this is rare when opioids are prescribed appropriately for suitable patients with chronic pain, and when treatment is properly monitored. Opioid treatment should be reviewed regularly and continued only if it is providing benefit with an acceptable side-effect profile.

All medicines should be taken according to the instructions provided by the doctor or the patient information leaflet.
Other medicines
Other medicines may be helpful to treat certain types of pain. These are sometimes called ‘adjuvants’ and they include certain medicines also used to treat epilepsy (pregabalin or gabapentin) and depression (e.g. amitriptyline).
Surgery

Surgical measures are used only as a last resort. They include:

- drainage of kidney or liver cysts (using a tube passed into the body, or directly by surgery)
- surgery to remove affected parts of the kidney (nephrectomy) or liver (hepatectomy)
- transplant of the kidney or liver.
Minimally invasive therapies

These are procedures in which the nerve supply to the body part causing pain is blocked by injections or removed by surgery. These procedures need specialist expertise.
Checklist: Managing pain

- Pain discussed and assessed (at each clinic visit)
- Advice given on self-care for pain
- Chronic pain investigated by referral to other specialists, as appropriate
- Options for pain management discussed and agreed
- Information about patient organisations and other forms of support

Notes and questions you would like to ask your healthcare team

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Here is a scale that you can use to record the severity of your pain.

0 1 2 3 4 5 6 7 8 9 10

No pain Moderate pain Unbearable pain
Wellbeing, personal and family life

This section explains how ADPKD can affect the wellbeing, personal and family lives of patients and their families, what can be done to help cope with these effects, and what support patients can expect.

Emotional impact >
ADPKD can have a significant emotional and psychological impact on patients and their families, and yet communicating about the disease can be difficult. Some healthcare professionals may not fully understand the impact ADPKD can have, both on physical and mental health.

Questionnaires > have been developed specifically to help measure the impact of ADPKD on quality of life and wellbeing.

Work >
Some patients with ADPKD find that the disease affects their ability to work.

Financial issues >
ADPKD can affect things like health and life insurance and mortgages.

What can patients and families do? >
There are many things that patients and families can do to help cope with ADPKD.

You're not alone!

What about children and young people?

Seeking professional help

Coming to terms with ADPKD
You’re not alone!

‘Contacting the self-help group was good for me. It was good to meet other affected people and relatives and exchange thoughts. It helped me to think positively again and not only see the negative things. As the wife of a man with ADPKD, I might have been more worried than my husband is himself. We can handle it better now – even if it is still a challenge every day.’
Isabel, Austria

‘It would be too much to say that the National Patient Day in Berlin saved my life, but at least it really changed the way I look at many things. Thank you so much!’
Sven, Germany

‘I was at a PKD meeting in Freiburg at the weekend. It helped me to see how many people are affected and to know that I am not alone.’
Patricia, Germany

‘Peer support is so important, be it from family, friends, support groups or medical staff. If you feel you need it, make sure to find it.’
Cathriona, Ireland

‘I am a transplant patient and my “personal psychologist” – Luisa Sternfeld Pavia, the Chairperson of the Associazione Italiana Rene Policistico (AIRP) patient organisation – has played a decisive role during the course of my disease. I thank her wholeheartedly!’
Roberto, Italy

‘It is truly important to feel sheltered by peers in patient’s organisations. In addition, by the doctors and nurses.’
José, Spain

‘If talking about ADPKD helps you, talk about it!’
Carmen, Spain

‘Often just hearing about other people’s experiences with PKD can make a difference. By joining the patient organisation on Facebook I now know there are people out there experiencing the same things as me, and learning from them really helps.’
Polly, UK
What about children and young people?

‘Announcing the disease to my daughter was difficult, as one is bound to feel guilty for being responsible for transmitting a genetic disease to your child. Luckily, she seems to be coping with limited anxiety. Also, the fact that she is working in the health field will certainly provide her with further tools to confront her disease constructively.’
Corinne, France

‘I found out I had a polycystic kidney when I was 40. I had three children, only one of whom is affected by the disease… At the beginning we chose not to say anything about the disease until they were 18 years old.’
Roberto, Italy

‘Avoid overprotection in teenagers, they are normal and healthy people even with ADPKD. They need information and education for the future, but it takes time to accept things and go to the doctor, it’s a personal decision the right time to cope with it.’
Ricardo, Spain
Seeking professional help

‘I was desperate after several years of dialysis and a donor kidney that did not work. Every day was a rainy day – sad and worrying. I’ve been trying to get professional support from a mental health clinic. Unfortunately, this has turned out to be very difficult.’

Claus, Austria

‘I have been getting psychological care for about 3 years, I already began to prepare for it before dialysis with regular meetings once a month. I think that was and is a very good “investment”. Neither your family nor your friends can “absorb” certain topics. It is important to discuss some things outside that context. The ADPKD self-help group in Nuremberg, Germany, is very supportive.’

Phillipp, Austria
Coming to terms with ADPKD

‘Once I realised how lucky I am to live these days, with the kind of healthcare and options available for dialysis and transplantation, I really get the feeling to be “happy” with my disease… I already lost friends and family to much more painful diseases or fateful incidents… At times I have had pain from surgery and other procedures, but compared with patients with liver, heart or lung diseases I have come to know via patient groups, I am really happy to “only” suffer from kidney disease. It has changed me as a person, also in a positive way. In ways, it has made me a more caring person – it taught me to have patience and appreciate some different values compared with other people my age.’

Claus, Austria

‘Don’t worry unduly about not being able to do your usual things. It’s a huge life event – create new usuals!’

Cathriona, Ireland

‘The motto of my life has always been “always positive”? Positivity helps to live life in the best possible way and reduces the perception of negative factors. Positivity reduces stress and improves life!’

Giovanni, Italy

‘It’s a choice. When you have a condition it’s a choice how you want to live with it. You can shut yourself off, be angry, or you can choose the other way.’

Brenda, Netherlands

‘You can worry about a lot of things and let worries overwhelm you, but it really only serves to worry about the things that you can really change or control yourself.’

Pedro, Spain

‘As patients with ADPKD, we learn to enjoy life despite our health.’

Salvador, Spain

‘It’s your body and you live with it. It was hard for me to accept that I was sick, but when you accept it, you have to respect it. Simply, I kept living my life.’

Anna, Sweden
Emotional impact

Worry and fear are common, following a diagnosis of ADPKD. Some people have difficulty coming to terms with the diagnosis and may wish to avoid thinking about it. Others may have a sense of relief that a diagnosis has finally been made, meaning that they can access care and support.

Some people have feelings of anger or resentment at inheriting ADPKD from a parent. It is important to remember that it is not the fault of the parents. The parents of people with ADPKD often experience feelings of guilt.

In the longer term, patients sometimes have feelings of helplessness and frustration, for example because of pain, body image issues, issues resulting from sexual problems, the effect of the disease on work and finances, the potential future need for dialysis, or concerns following a diagnosis of intracranial aneurysm. Some patients may experience depression related to their ADPKD.

Adolescent patients can face difficulties in coping with ADPKD while dealing with other pressures associated with physical and hormonal changes, education, relationships and family life.

The family members of patients can be affected by these issues, together with the impact of the disease on work and finances. Clearly, the diagnosis of ADPKD in a child has significant emotional and psychological effects on parents. Some parents may have difficulty deciding when, how and what to tell their children about ADPKD.
What can patients and families do?

Patients and families deal with the impact of ADPKD in many ways. General suggestions that may help include:

• Find a way to talk about problems – this may be done with a family member or friend, or professional help may be sought from a counsellor or other healthcare professional. It helps if you can fully explain how the disease affects you and your family.

• Some patients and carers may need care and support at times because of stress, depression or anxiety. You may wish to ask your healthcare team about the available support services, including counselling and mental health specialists. Early referral to this kind of support may help prevent more serious problems later.

• It can help to find out how other people affected by ADPKD feel about the disease and cope with it. Peer support from other patients and carers may be available locally in person, or online. Patient organisations, mental health charities and other support organisations can provide this kind of contact and information.

• It can be difficult to know what to say to children about ADPKD, and when it is right to do so. The UK PKD Charity offers specific advice to parents about talking to children about the disease. Genetic counsellors may also be able to help here.

Exercise and other types of physical and social activity may help to relieve stress.
Questionnaires

Questionnaires have been developed specifically to help measure the impact of ADPKD on quality of life and wellbeing. These include the ADPKD Genetic Psychosocial Risk Instrument (GPRI-ADPKD) (available in the ‘Supplementary data’ file here) and the ADPKD Impact Scale (ADPKD-IS).

In addition, a questionnaire called the Polycystic Liver Disease Questionnaire (PLD-Q) can help assess the effect of liver cyst symptoms. This can be found here (p. 11–16).

If you are interested you may wish to ask your healthcare team about these questionnaires.
In common with many other chronic and progressive diseases, ADPKD can affect employment for patients or carers who leave employment or limit their work to look after affected family members (e.g. spouses and children). Some people also experience difficulties at work because ADPKD limits the activities they can do or requires them to take time off for clinic appointments. Over time, many patients report a loss of personal and family earnings.

If you are medically assessed for employment your diagnosis of ADPKD might arise, especially if you give permission for the assessing company to access your medical records.

If ADPKD is causing problems with work, you might be able to:

- get advice on employment issues from social services (or adult social care) authority, citizen’s advice services and trade unions
- apply for social benefit payments to help you cope with financial difficulties – patients should discuss this with their social services authority
- get further advice on these matters from patient organisations in your country.

Having ADPKD can affect certain career choices – for example it may prevent you from joining the armed services. You may wish to seek advice from a careers advice service about this, especially if you are a young person with ADPKD.
Your diagnosis of ADPKD is likely to arise when you are assessed for health or life insurance, which can be a requirement for substantial financial loans such as mortgages. You may be asked to undergo a medical examination and you may also be asked about your family’s medical history, to check for inherited diseases. If you do not answer all questions honestly, your policy may be invalid when a claim is made. You should be sure to specify that you have ADPKD, as the prognosis of other forms of polycystic kidney disease may differ.

Certain types of insurance policies or mortgages may not be available for people with ADPKD, and available policies are usually more expensive than for people without the disease.

- Patient organisations, social services and consumer advice services may provide further advice on the local situation regarding financial issues and actions that patients and carers can take.
- Your healthcare team and patient organisations may also be able to provide information to help you communicate with financial services companies.
Checklist: Wellbeing, personal and family life

☐ Healthcare team assesses emotional and psychological impact of ADPKD
☐ Advice provided on self-help approaches for coping with emotional and psychological impact
☐ Referral to support services (e.g. counselling) and mental health services, where appropriate
☐ Careers advice available, where necessary
☐ Information about patient organisations and other forms of support

Notes and questions you would like to ask your healthcare team.

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Family planning
This section explains how ADPKD can affect family planning and pregnancy.

People with ADPKD can face difficult decisions when considering whether to have children. This is because pregnancy can be associated with certain risks in women with ADPKD and because their children might inherit the disease.

The EAF and PKD International believe that all patients with ADPKD should have access to family planning services, including counselling and advice on pregnancy, pre-implantation genetic diagnosis, and contraception. This includes young people and adolescents.

Sexual problems
Erection problems are common in men with chronic kidney disease. Women and men with ADPKD can also experience sexual problems related to body image issues and discomfort caused by kidney growth. You may wish to seek advice from your family doctor or nephrology team if you experience these problems.

Male fertility
Men with ADPKD may develop cysts in the seminal vesicles, but these do not usually affect the sperm or fertility.

Contraception
The female sex hormones, oestrogen and progesterone, may worsen liver cysts. Women with moderate-to-severe polycystic liver disease are generally advised to avoid the use of oral contraceptives containing these hormones. Other types of contraceptives are normally available.

Pregnancy
The majority of women with ADPKD have successful pregnancies. Pregnancy generally progresses normally in women who have ADPKD and whose blood pressure and kidney function are normal.

There is a higher risk of certain complications related to high blood pressure and reduced kidney function, and there are considerations regarding the use of some medicines.

Dialysis and transplantation
There are special considerations relating to family planning during dialysis and after kidney transplantation – you may wish to discuss these with your nephrology or dialysis team.

Pre-implantation and prenatal genetic diagnosis
ADPKD is caused by specific genetic mutations, which are usually inherited from a parent. A person with ADPKD faces a one in two (50%) chance of passing the disease onto each child they have.

Pre-implantation genetic diagnosis
Pre-implantation diagnosis (PGD) can show if an embryo created via in vitro fertilisation (IVF) has a genetic mutation linked to ADPKD. This allows people with ADPKD who want to have children to choose an embryo that does not have the ADPKD mutation, and therefore to prevent their children from having the disease.

PGD can only be performed if the specific genetic mutation causing ADPKD in the parent has been identified. More information about PGD is available here.

The EAF and PKD International believe that PGD should be available to all people with ADPKD, as recommended by the KDIGO Controversies Conference.

Results from one survey suggest that most people with ADPKD agree with this. However, access to PGD varies across Europe because of regulatory, ethical, legal and funding policies. Differing attitudes towards PGD can also affect access to this method.

Prenatal testing
It is possible to use prenatal testing to check whether an unborn baby in the womb has a genetic mutation linked to ADPKD. This usually involves testing small samples of tissue from the placenta, together with ultrasound scans.
Erection problems
Problems in gaining or maintaining erections (often known as erectile dysfunction or impotence) are most common in end-stage renal disease and have several possible causes. Kidney transplantation may not improve erectile problems in some men.
Complications

Women with ADPKD are more likely than women without the disease to develop high blood pressure and pre-eclampsia, a potentially serious complication involving high blood pressure and blood in the urine (proteinuria). This means that monitoring during pregnancy is particularly important for women with ADPKD.

Hormones released during pregnancy may increase the growth of liver cysts, but this is not likely to affect the liver function or the pregnancy.

The risk of infections of the kidneys and urinary tract are more common during pregnancy – these can be treated as necessary.

The growth of kidney cysts is not usually affected by pregnancy. However, having multiple pregnancies may increase the risk that kidney function declines over time.

Some experts recommend against pregnancy in women with moderate to severe chronic kidney disease (stage 3–5) caused by ADPKD because of risks to the mother and child.

Women with ADPKD may be advised to give birth in hospitals, rather than elsewhere, so that the midwife and obstetrician (a doctor who specialises in pregnancy, childbirth and the period after childbirth) can closely monitor the mother and baby.

The healthcare team, or a specialist obstetrician, will be able to explain these issues further and develop a monitoring and care plan for patients who wish to go ahead with a pregnancy.
According to a survey in the UK, just over half of 96 participating patients with ADPKD would have tried PGD (or might consider it in the future) if it were available on the National Health Service. The majority (69%) of patients believed that PGD should be offered to patients with ADPKD.
Medicines

Some medicines are not recommended for use in pregnant or breastfeeding women. For example, medicines called angiotensin converting enzyme (ACE) inhibitors or angiotensin receptor blockers (ARBs), used to control high blood pressure are not recommended for women who are pregnant or who wish to try for a pregnancy – your doctor may prescribe an alternative.
End-stage renal disease

This section explains end-stage renal disease (ESRD) and the treatment options for patients with ADPKD who reach this stage of kidney disease.

The kidneys have important roles that are essential to life. The kidneys of people with ADPKD often continue to work normally for many years. However, over time the growth and multiplication of cysts can interfere with the kidney function.

Eventually this can cause the kidneys to fail, meaning that they are not working well enough to support life. This is known as end-stage renal disease (ESRD), or stage 5 chronic kidney disease (CKD).

How common is ESRD in people with ADPKD?

Most people with ESRD need renal replacement therapy with either dialysis or kidney transplantation. Kidney failure is life-threatening unless it is treated by one of these approaches.

Doctors can predict the prognosis of ADPKD and estimate when a patient is likely to reach ESRD. This allows doctors and patients to discuss the available treatment options in advance and plan what to do if ESRD occurs. This is particularly important if a kidney transplant from a living donor is possible. Most hospitals have a special clinic to inform patients on the choices when they are approaching ESRD.

There are many diseases that can lead to ESRD. ADPKD is the most common inherited disease that can lead to dialysis and transplantation. Overall, around one in 10 patients with ESRD have ADPKD.

Local kidney and ADPKD patient organisations will have patients whom you can ask about ESRD and its treatment. Dialysis and transplantation are generally the same for all patients with ESRD and meeting fellow patients is usually a good and helpful experience.

Dialysis

Dialysis artificially filters the blood to remove the waste products and excess water that build up when the kidneys are not working.

Dialysis is used while patients with ESRD are waiting for a transplant, or if transplantation is not possible. Around nine out of 10 people with ESRD caused by ADPKD have dialysis as their first method of renal replacement therapy, i.e. before or instead of a transplant. Once dialysis is started, it must be continued for the rest of life, unless a transplant is performed. Dialysis only provides around 15% of normal kidney function.

There are two forms of dialysis: haemodialysis and peritoneal dialysis. Haemodialysis is used most commonly, but both methods are suitable for use in most people with ADPKD depending upon the individual circumstances. You may wish to discuss these options with your doctor.
Can I travel while on dialysis?

‘As far as I know, there are great dialysis holidays.’
Nora, Austria

‘We go on holiday every year and we have always had the opportunity to get dialysis. Of course, we always look in good time for a suitable centre. We have booked through dialysis holiday providers, but mostly we do it ourselves. We then send the doctor’s letter to the centre and go on vacation. When my husband still did peritoneal dialysis, we used to send the order so that the dialysis bags and accessories were delivered to our hotel before our arrival, without any problem. For us, dialysis is no reason not to enjoy our vacation.’
Valerie, Austria

‘Thankfully, it was possible to travel despite dialysis. In part, I was assisted by the dialysis team in my ward, but mostly I had to take care of getting in touch with dialysis centres myself. The payment was dealt with by the health insurance fund. I had to take care of the costs of travelling from the hotel to the dialysis centre and back,’
Claus, Austria
Dealing with dialysis

‘I would recommend everyone having dialysis to take something to do, read or play, it also helps to chat with your “neighbours” there, if they feel like it. If possible, you should try to keep busy, then time will pass by faster.’ Nora, Austria

‘For me it came suddenly “out of the blue” that I had to undergo dialysis. Thankfully, I had the opportunity to talk with some other dialysis patients and hear why they chose one or the other treatment method. I think that made it easier for me to accept my fate.’ Phillipp, Austria

‘When my partner started his dialysis treatment I felt relieved because until then his health continuously declined. The course of the disease turned an athletic and active father and partner into a tired, exhausted person. It was really painful to watch this without being able to help.’ Gabriele, Germany

‘We love dialysis, we don’t hate it. The treatment allows us to survive waiting for the transplant to improve our lives. It is true that we depend on a machine to survive, but at least you can find these machines everywhere around the world.’ Daniel, Spain

‘Being on dialysis was sometimes an emotional strain – and probably on my loved ones too. Waiting for a donor with no set date rather than having a live donor planned is just that – a “waiting game” – and it can be tortuous. But, however hard dialysis could get, I remembered it was keeping me alive.’ Andy, UK
Stages of chronic kidney disease

There are five stages of chronic kidney disease. These are defined by a person's estimated glomerular filtration rate (eGFR), a measure based on a blood test.

<table>
<thead>
<tr>
<th>Stage</th>
<th>Description of kidney function change</th>
<th>eGFR level (ml/min/1.73m²)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Normal kidney function</td>
<td>90 or higher</td>
</tr>
<tr>
<td>2</td>
<td>Mild loss</td>
<td>60–89</td>
</tr>
<tr>
<td>3a</td>
<td>Mild to moderate loss</td>
<td>45–59</td>
</tr>
<tr>
<td>3b</td>
<td>Moderate to severe loss</td>
<td>30–44</td>
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<tr>
<td>4</td>
<td>Severe loss</td>
<td>15–29</td>
</tr>
<tr>
<td>5</td>
<td>Kidney failure or end-stage renal disease</td>
<td>Less than 15</td>
</tr>
</tbody>
</table>
How common is ESRD in people with ADPKD?

Most people with ADPKD will reach ESRD at some point, but the age at which this occurs varies between patients. Around half of patients reach ESRD by the age of 60 years. Around two or three out of 10 people will never reach ESRD.
Haemodialysis uses a dialysis machine to filter the blood outside the body. Blood is removed via a needle in the arm and passed through a tube into the machine. Filtered blood is then returned to the arm via another tube.

**Preparation:** A small operation is performed to create a special blood vessel (called a ‘fistula’) in the arm, through which blood is passed to and from the dialysis machine. If there are problems with creating this fistula, a tube can be inserted into a large vein in the neck instead. This is called a ‘neck line’ and is used as a temporary measure.

**Procedure:** Haemodialysis is normally performed three times a week, with each session lasting around 4 hours. However, this can vary. Haemodialysis is performed at a nephrology clinic but in some cases it can be done at home. You can discuss your options with your clinic.

**Side effects and precautions:** People who have dialysis are instructed to limit the amount of fluid they drink. Special dietary requirements (normally advised by a dietician) are needed to reduce the intake of salt and other minerals – this helps reduce the build-up of fluid and minerals between dialysis sessions.

The main side effects include: tiredness, low blood pressure, sepsis (infection in the blood), muscle cramps and itchy skin. You can ask your healthcare team for further advice about these.
Peritoneal dialysis filters the blood through the lining of the patient’s own abdomen (called the peritoneum). A special dialysis fluid is instilled into the space in the abdomen (the peritoneal cavity). This fluid draws waste products and excess water from the blood as it passes through the peritoneum, and is then removed.

Peritoneal dialysis can be used instead of haemodialysis in many people with ADPKD. However, when the kidneys are very large or there are frequent cyst infections, peritoneal dialysis might not be a good option.

**Preparation:** An operation is performed to insert a permanent tube (called a catheter) through the skin into the abdomen.

**Procedure:** Dialysis fluid is pumped into the peritoneal cavity, left there for several hours and then drained into a bag. The process is then repeated, using fresh fluid, several times a day. If available, special machines can do all this overnight while you sleep. Peritoneal dialysis can be done by patients at home, but it must be done every day. CAPD allows you to do other things during fluid exchanges, e.g. use a computer, sew or watch television. A type of peritoneal dialysis called automated peritoneal dialysis uses a machine to do the exchanges overnight while you sleep.

The main side effects include: tiredness, peritonitis (infection of the peritoneum), hernia and weight gain. Some people find the catheter troublesome. You can ask your healthcare team for further advice on these issues.
Kidney transplantation

What is kidney transplantation?

Kidney transplantation involves removing a kidney from a donor and transferring it to a person with ESRD. People can live with just one kidney so only one donor kidney needs to be transplanted.

Where possible, pre-emptive kidney transplantation is performed before ESRD occurs. However, kidney transplantation is not suitable for everyone and the criteria for suitability can vary.

In rare cases, patients with severe liver cysts together with ESRD may be considered for a combined liver and kidney transplant.

Who donates the kidney?
The transplanted kidney can be provided by a living or deceased donor. You may wish to discuss these options with your nephrologist.

Living donor: A kidney donation from a suitable living person tends to work best and can be planned in advance as an ‘elective’ operation.

Deceased donor: The alternative is for patients to go onto a waiting list to receive a kidney from a person who has recently died.

Who donates the kidney?
The EAF and PKD International believe that patients with ESRD should be offered the opportunity to join a kidney transplant waiting list, if they are medically suitable.

What does transplant involve?

Kidney transplantation surgery involves a major operation organised by a transplant team and performed by a transplant surgeon.

While awaiting a transplant, it is important to stay as healthy as possible through basic ADPKD management and self-care measures. The transplant team should give specific instructions about what to do nearer the time of the operation.

Generally, transplantation is just as likely to be successful in people with ADPKD as with any other kind of kidney disease. As with any operation, there are risks and you should discuss these with the nephrologist or transplant surgeon.

Post-transplant care – what happens afterwards?

Most transplanted kidneys start to work immediately. Sometimes a transplant can take a few days or weeks to work properly, in which case dialysis may be needed temporarily. The time taken for recovery varies, but most patients should be able to leave hospital in around 1 week and to return to work and normal activities within a few months.

Long-term follow-up care is essential to make sure the transplanted kidney is working, to manage immunosuppressant medication necessary to prevent the body rejecting the new kidney, and to check for any complications.

Many core aspects of basic ADPKD management and self-care are still important, such as a healthy diet, weight control, smoking cessation, limiting alcohol intake and treatment for high blood pressure.

Remember, you will still have ADPKD even after a kidney transplant and so complications that occur elsewhere in the body may still need treatment.

Transplantation can have emotional and psychological effects both in patients and in donors. Anyone affected by these should discuss with their doctor the available options for advice, care and support.
Deciding about a transplant

‘I simply couldn’t ask anyone from my social circle to donate a kidney – I couldn’t have accepted it, although some friends did offer.’
Claus, Austria

‘Since I needed both a liver and kidney transplant, the decision for or against was very difficult. I decided to take the step only after a thorough preparation, which focused mainly on ethical and psychological concerns. My family was also an issue of course. My basic idea was first of all that the surgery could be fatal and second, that the transplantation and the possible lifetime thereafter would certainly be more successful if I made my decision independently of other people. Meanwhile, I’ve already experienced my first “transplant birthday”, with relatively few complications. That said, I still feel fragile sometimes.’
Selina, Austria

‘It was upsetting when strangers thought I was pregnant because of my enlarged liver… In a sense I feel emotionally more comfortable after the transplant. Previously I felt the “duty” of having to keep working and functioning until final kidney failure occurred – emotionally and mentally it was incredibly difficult and distressing. Carefree living was not possible, and apart from that, the responsibility towards my daughter, and the fear and panic not to be able to accompany her to adulthood in the worst-case scenario.’
Helena, Austria

‘After the doctors told me I needed a new kidney I was speechless. And that’s really rare for me!’
Petra, Germany

‘When my estimated glomerular filtration rate (eGFR) dropped below 20, it was suggested that I start thinking about live transplant. My great niece volunteered and was a really good match. From the time she agreed to donate to transplant was about 2¾ years. I had the transplant at eGFR 9, in October 2016. The transplant team told me that mine was a “textbook” pre-emptive live donation. My niece sailed through it – she wanted food as soon as she came back from theatre! The transplant was on the Thursday and she went home on the Monday. I was in the full week. Once out, I was very well looked after by my wife.’
Martin, UK
Life after a transplant

‘Even though I’m glad that dialysis treatment exists, I’m happy I don’t have to do it three times a week anymore – since I’m successfully transplanted now. Dialysis had taken away too much from my quality of life, though I managed to get it back it little by little. After many years with a transplant, I realise that one becomes careless and sloppy when it comes to taking immunosuppressant medicines. You know very well that you have to take them, but the “inner laziness” is sometimes bigger. Since you do not feel any pain or immediate reaction if you do something wrong, your attention decreases.’

Benjamin, Austria

‘The transplant operation went smoothly and I woke in recovery as if nothing had happened, apart from all the tubes. However, the following day I did feel nauseous, and was sick, then Day 2 I started to go into full recovery, and Day 3 the kidney started working and all tubes were removed. On Day 5 I was able to come home. Initially it went well. I have had a hiccups with a viral infection, but my lifestyle is getting back to normal. I’m feeling healthier, and about to start up physical fitness again. Even with the hurdles I’ve encountered, my GFR has been up to 65%, and I am looking forward to the future.’

Ian, UK

‘Dialysis hardly worked for me at all. The doctors had said that my need for a transplant was urgent. Around three months after an operation to remove my PKD kidneys, the consultant felt I could risk going on the transplant list. Just one week later, I was at home and I received a call to go immediately to the hospital. I had no reservations about having the transplant even though I realised I was vulnerable to infection and still had pain from recent surgery. My creatinine level went from 1,400 to 100 overnight and I woke up feeling like a brand-new woman. I was well cared for and a future felt possible once again. I have been on quite a journey over the last 13 years. Certainly, this donation prolonged my life span and increased my quality of life immensely – I have been able to travel, gain a Masters degree, continue to work for some time and create a home for myself. I have also had to deal with the traumatic impact ADPKD has had on my emotional and physical health, and have had some unfortunate experiences of employers and colleagues not understanding what this has been like. Even so I wouldn’t have had it any other way. To share life with another person is a great privilege in both directions.’

Nicki, UK

‘Immediately after the transplant, my skin lost its yellow tinge and I gained a normal complexion. I felt normal again, just like I used to. The biggest change was no longer being tied to dialysis three times a week, and once again being able to do all the things that I used to without having to take regular breaks. One thing I will never forget. How lucky I have been.’

Rob, UK

‘I have no dialysis and no dietary restrictions and my fitness is such that I play golf, have been back pedalling on the bike and I competed at this year’s Transplant Games. Life will never entirely return to what it was before, but every day feels like a massive bonus. I am immeasurably grateful to the medics and of course to my donor and his family.’

Stephen, UK
Pre-emptive kidney transplantation

Where possible, kidney transplantation is the best treatment for ESRD. This is because a successful transplant restores kidney function without the need for dialysis and can offer patients a better quality of life. It is also far more cost-effective for the health system, as compared with dialysis.

Preferably, kidney transplantation is best done ‘pre-emptively’ before ESRD occurs because this is most likely to be successful. A pre-emptive transplant allows patients to choose to receive a kidney either from a living donor or a deceased donor.
Criteria

Generally, patients must be healthy enough for surgery, have a good chance of a successful transplantation, and be willing and able to take certain treatments that are necessary after the transplant.
Living donor transplantation requires a suitable donor who is willing and able to undergo surgery to remove one of their kidneys. The donor is often a close relative as this reduces the risk that the patient’s immune system will attack (or ‘reject’) the donated kidney.

Potential donors are carefully checked to make sure they do not have ADPKD themselves – this may include genetic testing. Many other assessments are also recommended for potential donors, including tests for HIV, hepatitis B and C and cytomegalovirus (CMV) to reduce the risk that these infections are transmitted to the patient. Potential donors should also be counselled about the risks and requirements associated with donation.

Attitudes and policies regarding living donor transplantation vary across Europe. Living donor kidney transplantation is used most commonly in the Netherlands, followed by Turkey, Cyprus, Denmark and the United Kingdom. It is far less common in some other European countries.
Deceased donor

For a deceased donor transplant, patients need to go onto a waiting list to receive a kidney from a person who has recently died. Once a kidney is available, the transplantation operation is performed at short notice, as quickly as possible.

The length of waiting lists varies between countries. Patients may have to wait many months, or even several years, for a transplant. In the UK, for example, patients spend an average of 2–3 years on a waiting list. The main problem is a shortage of available kidneys. Patients should discuss the situation in their country. Once a deceased donor kidney becomes available, urgent transplant surgery is performed as soon as possible.
Kidney transplantation surgery

Kidney transplantation is a major surgical operation carried out under general anaesthetic. It may be done at a specialist transplant centre.

The abdomen is opened up and the donor kidney is inserted. The kidney is connected to the normal blood vessels and to the tubes that take urine to the bladder.

The patient’s own kidneys are usually left in place and normally these shrink after transplant surgery. However, one or both may be removed in certain situations, for example if they are very large and taking up space needed for the transplanted kidney, or if they are causing complications.
Risks

The risks of transplantation surgery include urinary tract infections, diverticulitis, blood clots and diabetes. There is also the risk that your body’s immune system might reject the new kidney. This risk is reduced using medicines that suppress the immune system, known as immunosuppressants.

Living kidney donors should also discuss the potential risks associated with donation, which include hypertension and increased costs of health insurance.
How long does a kidney transplant work for?

Many factors affect how long a transplanted kidney will continue to work and so this varies between patients. A large national study of patients with ADPKD in France showed that 93% of transplants were working after 5 years, 87% after 10 years and 79% after 15 years.

Patients whose transplant does stop working properly can normally go back onto a transplant waiting list.
Follow-up care

After a kidney transplant, long-term follow-up care is essential, for the following reasons:

• To check that the transplanted kidney is working and has not been rejected by the body. In the longer term, it is important to make sure that the kidney continues to work properly.

• To monitor immunosuppressant therapy and manage any side effects (depending on the specific drugs used); you should discuss these with your doctor.

• Importantly, by suppressing the immune system immunosuppressants leave you susceptible to infections. Simple precautions can be taken to avoid these, such as good personal hygiene, careful first aid for cuts and grazes, and avoiding people with known infections. You should contact your doctor if you are taking immunosuppressants and you think you might have an infection.

More information about living with a kidney transplant can be found here.
People who receive a kidney transplant usually need to take immunosuppressant medications for the rest of their life to prevent their immune system from attacking the new kidney. Commonly used immunosuppressants include tacrolimus, ciclosporin, azathioprine, mycophenolate, prednisolone and sirolimus.

Possible side effects of immunosuppressants include: diabetes, high blood pressure, weakening of the bones (osteopenia), changes in the cholesterol levels in the blood, and kidney problems. Importantly, these medicines can make patients vulnerable to infections. This means that precautions are necessary to reduce the risk of infections (such as vaccinations and good hygiene), and patients need to contact their healthcare team if they think they might have an infection. Symptoms of infection can include high temperature (fever 38°C/100.4°F or above), headache, aching muscles, diarrhoea and vomiting.
Follow-up care

This section explains the type of long-term follow-up care required by people with ADPKD.

If you have ADPKD you will need lifelong follow-up care involving a multidisciplinary care team, depending on your individual circumstances. How this care is co-ordinated depends on the local organisation of healthcare.

Nephrology care

Typically, if you have ADPKD you will remain under the care of a nephrologist. How often you see your nephrologist depends on many factors. These include your kidney function, your symptoms and complications, the type of treatment you receive and how quickly your disease is expected to progress.

Other specialists

The nephrologist will refer patients to other kinds of specialist doctors and healthcare staff if necessary, for example if they experience complications in other parts of the body.

Family doctors (also called primary care doctors or general practitioners) play an important role in providing and co-ordinating care for other diseases and aspects of health, as well as ADPKD.

What can patients do?

There are many ways that you can play a vital role in contributing to your own care. These include:

- Self-care
- Clinic appointments
- Prescribed medicines
- Monitoring and managing the effects of ADPKD
- Planning for end-stage renal disease

Remember that kidney and PKD patient organisations can be an important source of advice, support and information on these topics.

Transition care for adolescents

Adolescents with ADPKD face a transition from paediatric healthcare services to adult services.

Ideally, there should be a defined and co-ordinated pathway to transition for adolescents from paediatric to adult services, to help maintain continuity of care.
Factors

These include the level of kidney function, your symptoms and complications, and how quickly your disease is expected to progress. Clinic visits will also be needed more often for patients who start specific treatment to slow ADPKD progression.
Self-care

• Keeping up the recommended basic management and self-care measures is very important to help ensure you stay as healthy as possible.

• Ask your doctor or nurse for up-to-date information on ADPKD or visit patient organisation websites.
Clinic appointments

- Try to keep all appointments or reschedule in advance, as necessary.
- It will help your healthcare team if you give them as much information as possible about your health. For example, you should tell them about any changes to your health that have occurred, any specific symptoms or pain, any new medicines you are taking (prescribed or bought over the counter, including complementary therapies), and any side effects of treatment you have experienced.
- It might help to write down things you wish to tell the team in advance and take these notes to the appointment – perhaps using the checklists in this Route Map.
- Feel free to explain any effects of ADPKD on your wellbeing, personal and family life.
- Always ask questions if you are unsure about anything. It may help to prepare questions in advance. For example, three important questions you might like to ask are:
  - What are my options?
  - What are the possible benefits and risks of those options?
  - What help do I need to make my decision?
Some patient organisations provide help with asking questions.
Prescribed medicines

• Take all prescribed medicines according to the instructions given. Make sure you understand their possible side effects, any actions you can take to reduce the risk of these, and what to do if you experience side effects. You can ask your healthcare team about these issues.

• Tell your healthcare team about any changes to your health or the medicines you are taking. Contact your doctor, nurse or pharmacist if you have any questions or concerns about your medicines.
Monitoring and managing the effects of ADPKD

- It will help your healthcare team if you keep a record of your medicines, symptoms and general health.

- It can help to understand what stage of chronic kidney disease you have and what symptoms could indicate that this is getting worse. Patients in some countries have access to their own test results to help them monitor and manage their disease. You may wish to ask your doctor about these aspects.

- Make sure you understand the possible complications of ADPKD, what symptoms these can cause, what action to take and when.

- Agree with your healthcare team how you can best manage pain at home, and when you should contact the team.

- It can help to understand the roles and responsibilities of the various healthcare professionals involved in your care. Also, make sure everyone who treats you knows that you have ADPKD.

- Ask your healthcare team about the services available to help you deal with the impact of ADPKD on wellbeing, personal and family life.

- You can contact your healthcare team if you have any questions with respect to family planning and aspects such as the screening of family members for ADPKD.
If your ADPKD eventually progresses towards end-stage renal disease, it will be important to discuss with your nephrologist the available options for treatment, and to agree a plan.
Research

This section explains the types of ADPKD research in which patients may be able to participate.

Research is underway to help improve the scientific understanding of ADPKD and to improve care for patients. You may be able to participate in this research if you wish, depending on your circumstances and where you live.

Patients interested in participating in research should discuss this with their healthcare team, or contact their nearest kidney and ADPKD patient organisation or other specific research groups below.

Registries

Patient registries are databases that collect information about patients with specific diseases. This allows researchers to study various aspects of the disease, such as how it affects people, how it progresses over time, and how effective treatments are in practice. Registries are a valuable source of information, especially for rare or uncommon diseases such as ADPKD.

ADPKD registries exist in several European countries. There is also an international registry for children with ADPKD, called ADPedKD.

If you would like to join an ADPKD registry you should discuss this with your nephrologist. If you join, you will be asked to sign a consent form for the use of your information to be included in the registry.

Clinical trials

Clinical trials are research studies that test the efficacy (i.e. the ability to produce a desired or intended result) and safety of medicines or other types of treatment. Some clinical trials involve healthy volunteers, while others involve patients with specific diseases such as ADPKD.

There are several types of clinical trial. If you are interested in participating in this type of research, ask your nephrologist or contact one of the nephrologists on the list shown.

You can also find out more about clinical trials at the EU Clinical Trials Register, ClinicalTrials.gov and the PKD Foundation.

European reference networks

The European Reference Network for Rare Kidney Diseases (ERKNet) was launched in 2017 to promote high-quality, multidisciplinary care for rare kidney diseases, including ADPKD.

The ERKNet links together expert paediatric and adult nephrology centres in many European countries as you can see on this map – these centres are listed here.

These centres use uniform clinical guidelines and pathways, monitor the quality and outcomes of treatment, provide education for nephrologists, and support research. The network also offers virtual consultations for doctors who need advice and provides links to information for patients.

There is also a European Reference Network on Rare Liver Diseases (RARE-LIVER), including polycystic liver disease.
Scientific progress

‘I hope that scientific progress can improve both life expectancy and the course of treatment of all patients with polycystic kidney disease.’

Teresa, Italy
European countries
Countries in which ADPKD registries exist include Austria, Denmark, Finland, France, Germany, Greece, Italy, Netherlands, Romania, Spain, Sweden, Turkey and the United Kingdom.
Checklist: Research

☐ Available options for participation on the ADPKD registry explained and discussed, with agreement between the patient and doctors regarding participation

☐ Available options for clinical trials explained and discussed, with agreement between the patient and doctors regarding participation

Notes and questions you would like to ask your healthcare team.
Information for health policymakers and providers

This section provides recommendations to authorities responsible for healthcare policy, planning and provision at national and local levels.

The ADPKD Patient Route Map explains the main elements of good practice in ADPKD care to help patients, families, healthcare providers and policymakers work together to ensure everyone receives the care and support they need, at the right time. It can help healthcare providers and policymakers to design, adapt or assess co-ordinated services to efficiently address unmet needs among people affected by ADPKD, in the context of local conditions.

Unmet needs in ADPKD

ADPKD is a chronic, progressive, inherited disease that causes cysts to develop in the kidneys and which can affect many other parts of the body. Unmet needs in ADPKD include a lack of awareness among many health stakeholders and a lack of co-ordinated care pathways, resulting in significant variations in care. Individuals with ADPKD should have access to co-ordinated, patient-centred, multi-specialist care, as outlined in this Route Map and elsewhere.

Patient-centred care

All stakeholders, including national governments and healthcare providers, should support efforts to better inform patients and families and to empower them to act as fully informed and active partners in care. More about patient-centred care >.

Multidisciplinary care

Patients should have access to a nephrologist with expertise in ADPKD. Collaboration should be encouraged between the various specialists involved in ADPKD care to design and implement co-ordinated services. More about multidisciplinary care >.

Technology uptake

Advances in imaging >, genetic testing > and communications and information technology > could help to improve ADPKD care.

Transplantation

ADPKD is responsible for around one in 10 of all patients needing dialysis or transplantation. Kidney transplantation is the optimal treatment for kidney failure, providing excellent outcomes and being far more cost-effective than dialysis. Collaborative efforts are needed to improve access to transplantation in line with EU initiatives.

Conclusion

Collaboration between health policymakers, providers, professionals and patients is encouraged to design and implement co-ordinated ADPKD services and to promote awareness building, education and research.

Patients and families who wish to support or participate in ADPKD advocacy should contact the kidney or ADPKD patient organisation in their country, or PKD International.
Patient-centred care

All stakeholders, including national governments and healthcare providers, should support efforts to better inform patients and families and to empower them to act as fully informed and active partners in care. Patients should be supported to maintain self-care measures, to deal with the impact of the condition, and to participate in shared decision-making regarding healthcare policies, services and research.

This requires patients and carers to have access to accurate, up-to-date information about ADPKD, their own clinical data, and opportunities to participate in making decisions. Patients and carers should also have a clear understanding of the available services and how these can be navigated optimally. This Route Map is provided as a tool to help achieve this.
Patients should have access to a nephrologist with expertise in ADPKD. Collaboration should be encouraged between the various specialists involved in ADPKD care to design and implement co-ordinated services.

Where possible, a team approach with all specialties provided in one centre or clinic would be expected to benefit research, expert and patient networking, efficiency and patient outcomes. Where this is not possible, managed co-ordination and networking of local or national specialist services is important to optimise patient care. Managed co-ordination would be expected to facilitate prompt, accurate diagnosis, avoidance of duplication of tests, better management of disease complications and manifestations, evidence-based access to treatment to slow disease progression and ultimately to improve patient outcomes.

We encourage policymakers and providers to support the European Reference Network on Kidney Diseases (ERKnet) and European Reference Network on Rare Hepatological Diseases (ERN RARE-LIVER). These networks will facilitate the sharing of knowledge, experience, medical research, teaching, training and resources.
Information for health policymakers and providers

This section provides recommendations to authorities responsible for healthcare policy, planning and provision at national and local levels.

The ADPKD database shows that, over the last decades of growth, there has been an increase in the prevalence of ADPKD-related health problems, despite the existence of effective treatments. New automated imaging methods can reduce the costs and labour needed to measure total kidney volume (the most important marker of ADPKD progression). Increased uptake of these techniques would greatly benefit care.

Imaging

New automated imaging methods can reduce the costs and labour needed to measure total kidney volume (the most important marker of ADPKD progression). Increased uptake of these techniques would greatly benefit care.
Genetic testing

Access to genetic testing varies across Europe with key barriers including the cost of tests, resourcing of services, diverse reimbursement policies, and a lack of clear, reliable information in some countries. The EAF and PKD International believe that genetic testing should be available to patients for whom it is clinically indicated, and that all patients should have access to pre-implantation genetic diagnosis (PGD).

The uptake of faster and cheaper genetic tests could herald a greater role for genetic testing in the diagnosis of ADPKD and in predicting the disease prognosis.
Communications and information technology

New telecommunication and information technologies can facilitate multi-specialist networking, avoiding patients having to travel to access expert care that does not exist in their country. This technology can also promote patient empowerment and self-care.
### Patient organisations

#### Europe

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<th>Country</th>
<th>Organisation</th>
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<td>Belgium</td>
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#### Asia

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<th>Country</th>
<th>Organisation</th>
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<td>Japan</td>
<td>PKD Foundation</td>
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#### Australasia

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<tr>
<td>Australia</td>
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#### International

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<tr>
<td>Federation of European associations of patients affected by Renal Genetic diseases (FEDERG)</td>
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<tr>
<td>PKD International</td>
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Further reading

Many of these articles are freely available online.


Cnossen WR et al. Polycystic liver disease: an overview of pathogenesis, clinical manifestations and management. Orphanet J Rare Dis 2014;9:69


Edery MW. Male sexual dysfunction and chronic kidney disease. Front Med (Lausanne) 2017;4:32

European ADPKD Forum. Translating science into policy to improve ADPKD care in Europe, EAF, 2015 (Available here)


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This Route Map is published jointly by the European ADPKD Forum (EAF) and PKD International.

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Acknowledgments
The EAF and PKD International gratefully acknowledge review and input by:

Medical: Dr Nathalie Demoulin (Division of Nephrology, Cliniques Universitaires Saint-Luc, Université catholique de Louvain, Brussels, Belgium); Dr Esther Meijer (Department of Nephrology, University Medical Center Groningen, University of Groningen, Groningen, the Netherlands); Dr Lucas Bernts (Department of Gastroenterology and Hepatology, Radboud University, Nijmegen Medical Centre, Nijmegen, the Netherlands).

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Patients and families: all patients and family members who kindly provided the quotes included in this Route Map.

Sponsorship
Otsuka Pharmaceutical Europe Ltd initiated and facilitated the EAF and funded its activities. The ADPKD Patient Route Map and the EAF Multidisciplinary Position Statement were funded by Otsuka Pharmaceutical Europe Ltd and Ipsen Farmaceutica BV. No participants received fees in respect of this project. This Route Map represents the opinions of the authors and not necessarily those of the sponsors.

The authors acknowledge editorial support in the development of this Route Map by Interel (Brussels).