

# Autosomal Dominant Polycystic Kidney Disease

**Information Booklet for Patients, their Families and Carers**

**Published by:**

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## Preface

This booklet is for people affected by Autosomal Dominant Polycystic Kidney Disease (ADPKD), their family members, friends and carers.

Despite being twice as common as multiple sclerosis and affecting more people than cystic fibrosis, muscular dystrophy, haemophilia, Down syndrome and sickle anaemia combined, ADPKD remains the **silent disease**.

At present, there are no effective treatments for ADPKD. If the kidneys fail then management is by dialysis. Ultimately a kidney transplant may be necessary.

In this booklet, we describe the diagnosis, characteristics and management of ADPKD and explain the more common associated problems. It is not intended as a complete review but represents an overview of current information and opinion. As new research and knowledge becomes available some of the views expressed may be subject to change.

### *Please note:*

This booklet does not cover the other form of inherited PKD, autosomal recessive polycystic kidney disease (ARPKD). This more usually affects babies and young children, and requires different management to ADPKD.

For more information on ARPKD, please visit the PKD Charity website [www.pkdcharity.org.uk](http://www.pkdcharity.org.uk)

### **Disclaimer:**

The information provided in this booklet is not intended to be a substitute for advice provided by your doctor or other medical professional. Do not use this information for diagnosing a health problem or disease.

## Glossary of Abbreviations

ACE	Angiotensin Converting Enzyme
ADPKD	Autosomal Dominant Polycystic Kidney Disease
ARB	Angiotensin II Receptor Blocker
ARPKD	Autosomal Recessive Polycystic Kidney Disease
CAPD	Continuous Ambulatory Peritoneal Dialysis
CKD	Chronic Kidney Disease
CT	Computerised Tomography
CCPD	Continuous Cyclic Peritoneal Dialysis
ERF	Established Renal Failure
eGFR	Estimated Glomerular Filtration Rate
GFR	Glomerular Filtration Rate
MRA	Magnetic Resonance Angiogram
NICE	National Institute for Health and Clinical Excellence
RRT	Renal Replacement Treatment
UTI	Urinary Tract Infection

## Fast Facts about ADPKD

- Most common inherited kidney disease
- Characterised by presence of numerous fluid-filled cysts in the kidneys and often the liver and pancreas
- Affects between 1 in 800 and 1 in 1000 worldwide, ie at least 60,000 people in the UK, irrespective of gender and race
- Cysts increase in size and number over time displacing normal kidney tissue, impairing kidney function and sometimes leading to kidney failure
- Symptoms may not develop until 70% of kidney function is lost, typically in late 40s and 50s
- GFR – glomerular filtration rate - is best measure of kidney function
- Condition is highly variable even within families
- Not everyone with ADPKD will have kidney failure requiring either dialysis or transplant to survive
- ADPKD is incurable but progressive decline in kidney function may be reduced through preventative measures
- Blood pressure control is essential - target level is 130/80 mmHg (or 125/75 if you have proteinuria)
- Transplantation offers best outcome when kidneys completely fail
- Key factors in the progression of ADPKD are:

<b>Un-modifiable factors:</b> Kidney function as measured by GFR Inherited abnormal genes Gender, age Race	<b>Modifiable factors:</b> Blood pressure Diet Smoking Exercise
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## What is Autosomal Dominant Polycystic Kidney Disease (ADPKD)?

### The world's most common inherited kidney disease

In ADPKD, numerous fluid-filled cysts develop from the **tubules** of the kidneys that carry urine away. Over time, the cysts grow and multiply causing the kidneys to function less effectively and often fail. The cysts gradually replace healthy tissue causing the kidneys to enlarge, often 3 to 4 times normal size. ADPKD is a multi-system disorder and patients may also experience complications such as:

- hypertension (high blood pressure, a common feature of the disease)
- lower back pain (common)
- blood in the urine (common)
- abnormalities of the heart valves (common)
- liver cysts (common)
- pancreatic cysts (uncommon)
- brain aneurysms (uncommon)

ADPKD is defined by the Department of Health (DH) as a "**long-term, progressive, chronic kidney disease (CKD)**"<sup>1</sup>. It is incurable and affects men, women, and races equally. It is typically diagnosed between the ages of 30 and 50 years.

In the early stages of ADPKD, there are usually no symptoms, and it may be undetected until a patient is found to have some of the conditions listed above or through family screening. Once diagnosed, it is essential to measure kidney function regularly and treat the symptoms and complications so as to combat the adverse risks to health of cardiovascular disease and renal failure.

ADPKD is clinically very variable even within families. Not everyone will develop complete renal failure – known as **Established Renal Failure** or **ERF** requiring either dialysis or transplantation.

No matter the age you are diagnosed it is not possible to predict the severity or range of medical problems you may have. For instance, about 50% of people over 60 with ADPKD will have ERF but one third will get to 70 without evidence of failing kidneys and live a normal life. About 60% of patients, however, will develop high blood pressure (BP), which can be treated with medication.

### How do you get ADPKD?

ADPKD is almost always inherited<sup>2</sup>. A **hereditary disease** is one that is passed on from a parent to a child through their genes.

Every person has **23 pairs of chromosomes**, which contain the genetic information called **genes**. One of the 23 pairs of chromosomes is the 'sex chromosomes' which are involved in determining gender. The remaining 22 pairs of chromosomes are 'autosomal' and contain all the other genes. Since ADPKD is **autosomal** it affects males and females equally.

There are two genes that can be mutated to produce ADPKD. The most common form (85% of cases), **PKD-1**, results from changes on a gene on chromosome 16. The remaining 15%, **PKD-2**, are caused by mutations in a gene on chromosome 4. PKD-2 is believed to be less clinically severe than PKD-1.

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<sup>1</sup> See the end of the booklet for more on the **5 Stages of CKD**

<sup>2</sup> New genetic mutations occur in about 4% of ADPKD patients. Cysts mimicking PKD may also occur as non-inheritable changes as a result of other long-term kidney problems, dialysis and old age.

ADPKD is termed **dominant** because an abnormality in one of the pair of genes will produce the disease. In other words, an affected individual has one normal and one mutated gene, and as each parent passes on one of each pair of their genes, i.e. half of their genes, an affected parent has a 50 % chance of passing down the abnormal gene to any child they might have.

ADPKD does not skip generations; and so if a child does not inherit the abnormal gene they will have normal kidneys and likewise cannot pass on the condition any further.

Although ADPKD has been identified as an inherited disease since the 1920s, the genes for PKD1 and PKD2 were only identified in 1994 and 1996 respectively. Substantial research is being undertaken to understand how these genes cause ADPKD and also find treatments to prevent the growth of cysts.

### **How is ADPKD diagnosed?**

Up to 70% of kidney function may be lost before symptoms develop. So ADPKD is typically diagnosed in adulthood as it is frequently without symptoms until mid-life. However it can more rarely be diagnosed in about 2% of babies where there is a family history of ADPKD.

A family history of ADPKD may lead to an early diagnosis through screening. Sometimes diagnosis may follow abnormalities noticed during a routine medical or a pregnancy scan. Normally, however, diagnosis follows after a visit to the GP with symptoms such as:

- Back pain on either side of the spine
- High blood pressure (hypertension),<sup>3</sup>
- Blood in the urine (haematuria)
- Repeated urinary tract infections
- Infection in a kidney (pyelonephritis)
- Renal colic from kidney stones (Nephrolithiasis)
- Persistent headache
- Swelling of the ankles
- Loss of appetite

***More information on these symptoms is given later in this booklet.***

The process of diagnosing ADPKD includes:

- Taking a full personal and family history
- Carrying out a thorough physical examination and examining the abdomen for any abnormalities such as large kidneys
- Measuring **blood pressure**
- **Ultrasound** scanning of the kidneys - using sound waves (not x-rays) to "see" the kidneys (similar to how babies are seen in the womb during pregnancy)
- Blood and urine tests to look at chemical composition, in particular levels of **creatinine**
- Sometimes, a CT (computerised tomography) scan may be required for a more detailed picture of the kidneys
- If there is a history of **aneurysms** (enlarged blood vessels in the brain), an MRI scan may be arranged

The standard diagnostic criteria for ADPKD are:

- At least 2 cysts in 1 kidney or 1 cyst in each kidney in an at-risk patient under 30 years

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<sup>3</sup> A full explanation of blood pressure measurements is provided later in the booklet

- At least 2 cysts in each kidney in an at-risk patient between 30 and 59 years
- At least 4 cysts in each kidney for an at-risk patient over 60 years
- Diagnosis is supported by the presence of hepatic (liver) or pancreatic cysts

If cysts are present on the kidneys, they are usually easy to see on a scan. But remember that cysts develop with age, and until the age of 30, the absence of cysts does not necessarily mean that the person has not inherited the condition. For this reason, people under 30, who have family history of ADPKD, should not be entered into kidney donor programmes.

As indicated, the scan may also show cysts on other organs, such as the liver and pancreas. While these organs will remain **functionally unaffected**, the cysts themselves may cause discomfort from pressure.

### **Who needs to know about a diagnosis of ADPKD?**

It is not essential to tell an employer, bank or building society, or insurance company<sup>4</sup>, unless a medical history is requested. However, if symptoms result in extended absences from work, it may be advisable to explain the reasons to an employer.

When applying for life insurance or critical illness cover, all insurance companies now ask whether ADPKD runs in the family and may want to investigate further before providing cover. It is often difficult and can be expensive to obtain these types of insurance policies, if you have been diagnosed with ADPKD.

Kidney specialists (nephrologists) and other consultants will provide a medical report but only with the patient's permission.

Holders of HGV or PSV driving licences must tell the DVLC of the diagnosis when providing details of their medical history.

We do recommend openness with current or potential spouses/ partners and children.

### **How does ADPKD affect the kidneys?**

It is helpful to understand how normal kidneys function before looking at the effects of ADPKD.

#### **The normal kidney**

There are two kidneys in the body. Bean-shaped and normally about 11cm (4") in length and 150g (5oz) in weight, they are at the back of the abdomen on either side of the spine (Figure 1).

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<sup>4</sup> The government has imposed restrictions on the use of genetic test results by UK insurers until November 2011.

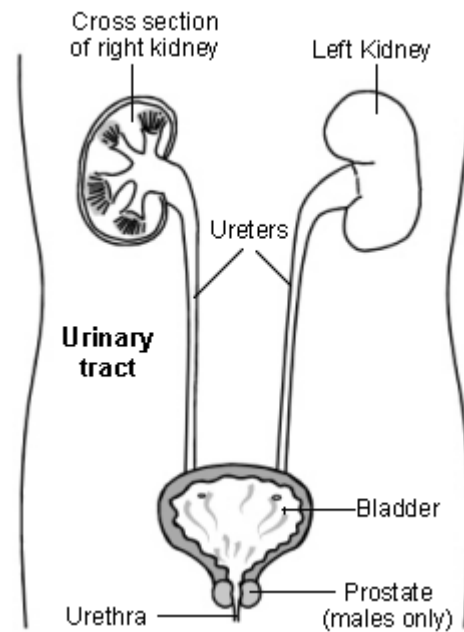


Figure 1: Location of the kidneys  
(Drawing courtesy of Patient UK)

The kidneys act mainly as a **filter**, purifying the blood of waste products and regulating the balance of salts and water in the body.

Each kidney contains approximately one million individual kidney filter units called **nephrons**. Each nephron is composed of a 5cm (2") **tubule** with its own small blood vessel. Blood containing waste products from the body passes through filters in the nephron called **glomeruli**, which hold back blood cells and protein, while allowing water, chemicals and salts into the tubules.

The tubules process this fluid and the excess flows as urine into the ureter and onto the bladder (Figure 2). Measuring the levels of the various waste products in blood and urine therefore can help assess kidney function.

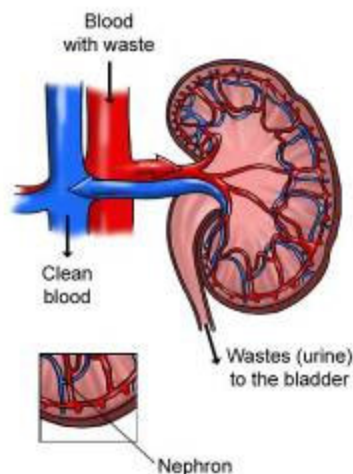


Figure 2: How the kidney works

The kidneys also produce three important **hormones** (Figure 3).

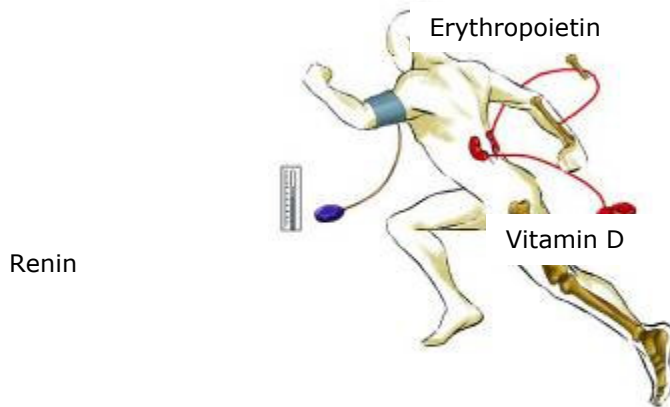


Figure 3: The three hormones produced by the kidneys

- **Renin** helps to regulate blood pressure and the balance of salt in the body
- **Erythropoietin** stimulates the bone marrow to produce new red blood cells
- **Vitamin D** is needed for calcium absorption and important for maintaining healthy bones

### **The polycystic kidney**

In the polycystic kidney, cysts first develop in parts of the tubules but over time hundreds of cysts form throughout the kidney. The nephrons eventually become compressed and distorted, and less and less effective at processing waste products.

Cysts can grow to 2cm in size causing the kidney enlargement common in ADPKD (Figures 4a and 4b). Severely damaged polycystic kidneys can weigh up to 17kg (38lb) each.

Figures 4a and 4b show the difference between a normal and a polycystic kidney at an advanced stage of ADPKD.



Figure 4a: A polycystic kidney in an advanced stage of ADPKD compared with a normal kidney  
(Photo courtesy of the PKD Foundation)

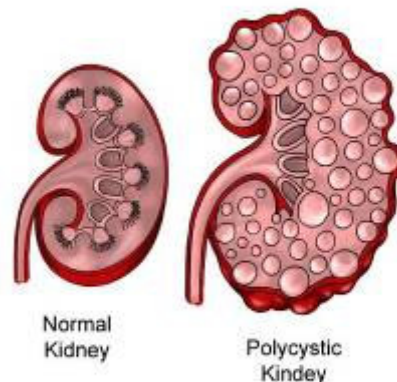


Figure 4b: Cross section of normal kidney compared with polycystic kidney

### **How is kidney function measured?**

As the kidney is a filter, kidney function is measured by its **filtering capacity**.

The usual method is to measure the level of **creatinine** in the blood. Creatinine is a waste product of muscle activity and it should remain constant over time if kidney function is normal. As kidney function declines, creatinine levels increase and it has therefore become a proxy for kidney function.

However, creatinine can vary from person to person due to differences in factors such as protein intake, muscle mass, gender and race – all of which have nothing to do with kidney function.

Blood creatinine levels rise only when the kidney has lost almost 50% of its reserve capacity and many now see a rising creatinine as a late indicator of kidney function.

**Currently, the standard method of assessing kidney function is to use an estimate of the GFR or glomerular filtration rate.**

**GFR is the rate at which the glomeruli in the kidneys excrete waste products and excess fluid. It reflects the percentage of normal filtration function remaining.**

**GFR is measured as mls/min, i.e. the amount of fluid (mls) filtered per minute (min). The GFR ranges from 0 to 100 mls/min and can be used as an equivalent indicator of kidney function from 0-100%.**

**The estimated GFR or eGFR is calculated from blood creatinine using formulae that take account of factors such as age, race and gender.**

**For more on eGFR and an online calculator to find your own eGFR, visit the Renal Association website:**

**<http://www.renal.org>**

**However, please be aware that this is only an estimate and not an accurate measure of your true kidney function.**

ADPKD patients should ask their renal consultant or nephrologist for their eGFR or request, as eGFR is used by the Department of Health to identify a patient's stage of chronic kidney disease (CKD).

When GFR falls below 30 mls/min, kidney function is considered to be 'severely impaired' and when GFR is below 15, the patient requires dialysis or transplantation.

### **Decreased kidney function and established renal failure**

The most important functional abnormality of ADPKD is reduced kidney function and, sometimes, established renal failure (ERF).

As the size and number of kidney cysts progressively increase, normal kidney function is reduced. The rate of deterioration varies and can take many years. It does not happen suddenly. Some ADPKD patients with impaired kidney function can live a normal life, without dialysis, despite up to 80% of kidney function loss, ie a GFR of no less than 20mls/min.

On average, however, approximately **50% of ADPKD patients over 60** will have ERF but ERF varies between 2 and 80 years, even within families.

However, when kidney function is very poor, waste products build up and this leads to symptoms of ERF. At this stage renal replacement treatment (RRT), either dialysis or transplantation is usually required.

It is important to emphasise that it is virtually impossible to predict who will develop ERF and when – family history is not a useful indicator. However, frequent monitoring of kidney function can help determine the rate of progression of the disease.

### **Hypertension**

Hypertension or high blood pressure (BP) is both a consequence and a contributor to kidney failure. At least **60%** of people with ADPKD have high BP. The reason for the rise

in BP is unknown, but it should be treated early. A sustained rise in blood pressure leads to thickening of the heart muscle, which can lead to a greater risk of heart disease, strokes and atheroma (hardening of the arteries). High BP also puts extra strain on the kidneys and can accelerate kidney function decline.

Where appropriate, patients will be advised to reduce weight, cut down on salt intake, moderate alcohol intake and stop smoking. Often, however, medication is required to reduce blood pressure levels.

See the section later on treating high BP.

### **Haematuria (blood in the urine)**

Around **50%** of people with ADPKD will get blood in their urine. The urine may be red, brown or smoky colour when there is blood in it. It may be easily seen or be so slight that it is only picked up on special urine stick testing.

Haematuria may last for several days, or rarely for many weeks, but invariably clears up on its own. Treatment is seldom required. The cause is thought to be damage to the kidney's small blood vessels caused by the cysts increasing in size. Sometimes the kidneys' delicate blood vessels may bleed directly into a cyst. This can be associated with pain and fever.

### **Kidney stones**

Kidney stones occur in about **20-25%** of patients with ADPKD. The reasons for this are not clear. Kidney stones are made up from various substances found in the urine. The sizes of stones vary from tiny particles to large, smooth or irregular lumps. The symptoms will depend very much on the size and position of the stones. Occasionally small stones may be passed without problems or discomfort, but larger stones may cause kidney obstruction and/or severe pain.

### **Back pain from large kidneys**

Polycystic kidneys can grow to 15cm (6") in length. In some patients, they cause a dragging feeling on either side of the abdomen. Some patients can feel their own kidneys with their hands. However, the kidneys themselves cannot directly damage other organs, although cysts do occur in other organs (such as the liver).

## The Effects of ADPKD on Other Organs

### Intracranial aneurysms (ICA)

10% of ADPKD patients are at risk of developing an ICA or berry aneurysm. This is an out-pouching of a blood vessel wall and most commonly happens in the larger blood vessels of the brain. See Figure 5.

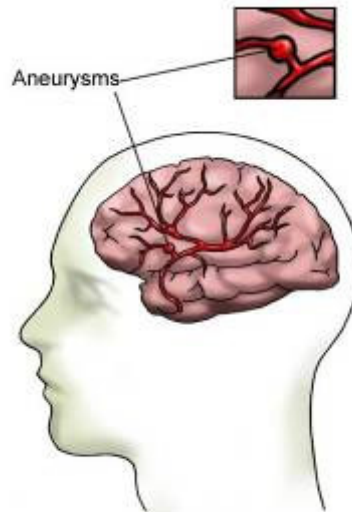


Figure 5: Aneurysms in the brain

These pouches or ICA can burst without warning, causing strokes. Typical features of an ICA rupture are a sudden, intense, unusual headache, often described as an explosion inside the head. Other symptoms include nausea and vomiting, an abnormal sensitivity to light, double vision, sleepiness and even transient loss of consciousness.

High blood pressure puts more force on the wall of an aneurysm and may make it more likely to leak or burst, so control of blood pressure is crucial.

ICAs occur about twice as commonly in patients where another family member is known to have had one. People at risk are recommended to have a magnetic resonance angiogram (MRA) every five years. If an aneurysm is identified, your specialist will advise you about the best management. Different surgical techniques (some of which do not need an open operation) can be undertaken to prevent such ICA bleeding at a later date.

### Liver cysts

The liver is the most common organ other than the kidney in which cysts are seen. It is affected in 40-70% of people with ADPKD. However, cysts are more common in women than men and tend to occur at a younger age.

Very rarely, a patient may develop massive hepatic liver disease. This is usually seen in women and is associated with oestrogen intake.

Fortunately the cysts rarely, if ever, cause any liver problems though they may sometimes cause pain from pressure effects. If the liver becomes exceptionally large, surgery may be necessary to relieve the compression of other organs; for example, breathing can be affected if the size of the liver presses up on the diaphragm.

### **Urinary tract infections (UTI)**

Women often have urine infections in the bladder, called cystitis, even though they may not have polycystic kidney disease. Any urine infection in a man and more than 3 attacks of cystitis in a woman deserves investigation in its own right.

For patients with a family history of polycystic kidney disease, these investigations should be requested sooner. Infection usually starts in the bladder but may pass up to the kidneys. More rarely, infection from the bladder or the kidney may get into the cyst fluid itself. Both types of infection can be treated with antibiotics.

### **Mitral valve prolapse**

In 20% of patients, a minor heart murmur can be detected. This is caused by the leaking of blood from one of the heart valves (usually the mitral valve). It does not usually get worse, but it is important to protect the valve from infection by taking antibiotics just before any dental procedure or major surgery.

### **Anaemia**

The kidneys produce a hormone called **erythropoietin** which stimulates red blood cell production from bone marrow. When GFR falls below 60, erythropoietin levels may decline, leading to anaemia. Symptoms include fatigue, lack of vitality and shortness of breath on exertion.

However, in ADPKD, anaemia develops later than with other forms of kidney disease because erythropoietin levels are better preserved.

### **Bone disease (hyperparathyroidism)**

Bone disease is rare but may be diagnosed in ADPKD where GFR is below 60. Reduced kidney function results in lower calcium and higher phosphate which can weaken bones and cause fractures. Skin itching is also a symptom of high phosphate.

Treatments include reducing phosphate in the diet (milk, cheese, eggs, chocolate) or taking phosphate binders with meals.

### **Diverticular disease**

The small outpouchings of the large bowel (colon) known as diverticula are more common in patients with ADPKD than the general population. Symptoms are rare unless they get infected or burst, when the patient experiences something resembling appendicitis but on the left side.

## Genetic Testing and ADPKD

### Is genetic testing useful for ADPKD and is it available in the UK?

At present, for the majority of patients, testing and diagnosis can be effectively achieved by clinical examination and scanning, as described above. It is not necessary to use genetic or molecular testing - which involves looking at blood DNA for an abnormality that may indicate a predisposition to a disorder, or confirms a suspected mutation.

If ADPKD is diagnosed in the family, other adults may wish to be screened, but this is not essential if they have no symptoms. It is always advisable to check blood pressure though.

It is not generally necessary to screen children under 20.

It is possible to request a genetic test for PKD-1 and PKD-2 through a Regional Clinical Genetics Centre<sup>5</sup>, for example where there is a significant risk to an unborn child.

Similarly, if a relative of someone with PKD is considering donating a kidney and scans are not conclusive, a genetic test may help the donor decide.

Genetic Centres provide information and help for NHS patients and their families who require genetic advice, diagnosis and management.

Making a diagnosis of ADPKD will enable regular monitoring and allow early and appropriate treatment of complications such as high blood pressure and urine infections. But the fact of knowing that you have ADPKD, with its potential threat to health and life in later years, may cause psychological difficulties, problems with obtaining life, critical illness and travel insurance or with employment in some occupations (eg Armed Forces).

### Pregnancy and ADPKD

Having a child may precipitate the question of genetic testing. If ADPKD is present in one parent, there is a 50%, 1 in 2 chance, that each child will inherit the same abnormal gene. Conversely, there is a 1 in 2 chance the child will not.

Parents must ultimately decide for themselves on whether to undertake genetic testing. Wider issues such as how ADPKD has affected the parent, complications in other members of the individual's family, age at which a couple decide to have a family may all impact on the decision to test in pregnancy.

As a general rule, however, antenatal diagnosis is not recommended except in rare families where a severely affected baby has been born.

Pregnancy itself does not necessarily affect the progression of ADPKD and generally women with ADPKD have normal pregnancies. However, there is an increased risk of developing urine infections and high blood pressure during pregnancy that will put an extra strain on the kidneys. It is therefore important to ensure your doctor and obstetrician are aware of your condition so that a close eye can be kept on your blood pressure and general health.

Ask your GP for a referral to a clinical geneticist to discuss further.

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<sup>5</sup> See the following websites for further information on UK Genetics Centres:  
British Society for Human Genetics <http://www.bshg.org.uk/>  
Dept of Health website <http://www.doh.gov.uk/genetics/gtn.htm>

### **Testing children for ADPKD**

At birth, very few children with ADPKD have any problems. Modern scanning techniques are sufficiently sensitive to see kidney cysts in children from families known to have ADPKD, but these may not necessarily be associated with clinical symptoms or poor kidney function<sup>6</sup>.

As outlined earlier, an early diagnosis of ADPKD can have disadvantages. Deciding whether your child is screened, therefore, raises important ethical issues and consequences. Furthermore, the child's views need to be taken into account. A visit to a genetic centre may be helpful here; and children who already know they have ADPKD may equally benefit from discussing their concerns in a genetics clinic.

Of course, if any child develops high blood pressure or urinary tract infections, scanning is often recommended and they will require prompt treatment to prevent complications and reduce the risk of kidney damage.

### **Clinical Geneticists**

Referral to a clinical geneticist can help you get a better understanding of the hereditary nature of ADPKD, if you are affected or at risk of the disorder. Geneticists will give information about diagnosis, inheritance and risks to the individual and the family, and potential treatment. Psychological counselling is also available.

Not everyone will require genetic advice or counselling but at some stage in their lives, people with ADPKD may find it particularly helpful. These situations may include finding out more information on the disease, considering the future, planning a family, or seeking advice about how to tell children that they may have inherited the condition. When specialist advice is required a GP should make a referral to a clinical geneticist at a suitable hospital.

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<sup>6</sup> Cysts may be observed on normal kidneys.

## What can be done to treat ADPKD?

There is no cure for ADPKD as yet. Management of the disease centres on prevention of complications, such as high blood pressure and infections, to delay loss of kidney function. It is possible to lead a normal life despite ADPKD but it is wise to be referred to a kidney specialist early.

If there are no complications and blood pressure is well controlled, appointments need only be once every year. Blood pressure and specific tests of kidney function need to be carried out annually. Complications such as urine infections, large amounts of blood in the urine or poor blood pressure control, require medical attention and prompt treatment.

### Controlling blood pressure brings benefits

High blood pressure (BP) is a symptom of ADPKD and has been found to accelerate the progression of kidney disease generally.

Moreover, the higher the BP, the greater the risk of cardiovascular disease such as stroke, heart attack and heart failure.

Recent large-scale studies in the US have demonstrated the benefits of **aggressive blood pressure control** on kidney function, so it's critical to keep blood pressure at normal levels at all times.

High BP rarely has symptoms. The only way to know if BP is high is to have it measured.

Blood pressure is the pressure of blood in the arteries. Every time the heart pumps it forces blood through the arteries and into smaller blood vessels called capillaries. The force produced by the heart as it pumps is called blood pressure. When the heart contracts and forces blood through the arteries, blood pressure goes up, when the heart relaxes it goes down<sup>7</sup>.

### BP is measured in 'mmHg<sup>8</sup>' by two numbers:

- The top number or **systolic**, eg 130, measures the pressure when the heart contracts or beats
- The bottom number or **diastolic**, eg 80, measures the pressure in between beats when the heart rests

The actual numbers vary between individuals and change with age and activity. Consistent BP readings over 140/90 are considered hypertensive. See Figure 6 below.

Normal levels for BP in the UK are 140/85, but ADPKD patients are recommended to keep BP at **130/80** or **125/75** if you have proteinuria (excessive amounts of protein in the urine).

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<sup>7</sup> Visit the The Blood Pressure Association charity website [www.bpassoc.org.uk](http://www.bpassoc.org.uk) for more on blood pressure

<sup>8</sup> The numbers represent the height in millimetres that mercury (Hg) rises in a column in line with heart beat – often translated into a digital readout

Category	Systolic (mmHg)		Diastolic (mmHg)	Meaning
Normal	Less than 120	And	Less than 80	Excellent
Pre-hypertension	120-139	Or	80-89	Your BP could be a problem
Hypertension	140 or higher	Or	90 or higher	See a doctor

Figure 6: What blood pressure readings indicate in adults

Some **non-medication techniques** alone can sometimes reduce high BP. These include:

- Maintaining an ideal weight
- Taking regular aerobic exercise – but avoid contact sports, such as rugby, and activities that could result in trauma to the kidneys and cause cysts to bleed
- General healthy eating – but see section below on special dietary factors
- Reduced salt - no more than 2.4mg Na (sodium) per day
- No smoking
- Reduced alcohol intake - maximum 1 unit/day for women; 2 units/day for men

Medication needs to be prescribed, however, where the above measures are not sufficient to control blood pressure.

A number of **drugs** are used to control high blood pressure and manage hypertension<sup>9</sup>. These include:

- **ACE inhibitors** (few side-effects, a cough being the commonest) or **ARB** (angiotensin II receptor blockers)
- **Diuretics**
- **Beta blockers** or **Calcium Channel Blockers**

ACE/ARB medications have been shown to reduce the rate of kidney function decline in several kidney diseases. In rare cases, some patients show a dramatic fall in their kidney function with the use of ACE/ARB drugs and medication needs to be stopped. It is therefore important that kidney function is checked regularly after starting these tablets.

### Dietary factors to consider with ADPKD

While kidney function is above a **GFR of 60**, the recommended diet is one based on the **DASH Eating Plan**<sup>10</sup> which was developed in the US and is clinically proven to significantly reduce BP. In summary, this specifies:

<sup>9</sup> See the NICE website for Guidelines on the Management of Hypertension in Primary Care

<sup>10</sup> DASH = Dietary Approaches to Stop Hypertension

Visit [www.pkdcharity.org.uk](http://www.pkdcharity.org.uk) for a copy of the DASH Eating Plan.

- Low-fat dairy products
- Fish
- Low levels of animal protein, eg poultry – 0.8g per kg body weight per day, ie around 50g for an average woman
- Fresh fruit
- Fresh vegetables
- Nuts
- Whole grains
- Generous intake of calcium, potassium, magnesium, fibre
- Reduced intake of salt/sodium, red meat, cholesterol, sugar

Figure 7 illustrates a balanced diet.



Figure 7: Balanced diet

However, when GFR falls below 60 and for patients with ERF, the DASH diet should be avoided as it can be too high in **potassium, protein and phosphate**.

There is limited or no evidence to support replacing animal protein with soy.

### Drink adequate quantities of fluid

Aim to drink at least 8 to 10 glasses of fluid a day, half of which is water. However, excess fluid may be a problem if diuretics are being taken.

If undertaking extreme exertion such as intense running, cycling or work, more fluid will be needed to prevent dehydration and reduce the risk of complications.

### Cyst removal is not recommended

There is no evidence that cyst removal or reduction prevents kidney function decline. This is also an impractical procedure because there are too many cysts and they continue to occur throughout life. Some cysts can also be extremely small.

However, a particularly large cyst that is causing pain or repeated infection may require drainage by needle in the x-ray department. Such large cysts in the kidney or liver can sometimes be "stuck down" to prevent them re-filling.

## Dialysis

When GFR falls below 30, there is increased risk of progressing to renal failure. At that point, either dialysis or transplant will need to be considered.

The dialysis options are:

**Haemodialysis:** a procedure that removes extra fluid, electrolytes and wastes from the blood using a dialysis machine either at home or at a dialysis centre, 3 times a week. Each session can take around 4 hours to complete during which time blood is pumped out of the arm, filtered through the machine and put back into the arm. Fluid intake and diet are usually strict, and anaemia can occur.

**Peritoneal dialysis:** this procedure involves using a soft tube (catheter) to fill the abdomen with a cleansing liquid called dialysis solution. The walls of the abdominal cavity are lined with a membrane called the peritoneum, which allows waste products and extra fluid to pass from blood vessels into the dialysis solution for subsequent drainage. The waste solution is poured away. Each process of filling and draining is called an 'exchange'. There are two types of peritoneal dialysis:

- Continuous ambulatory peritoneal dialysis (**CAPD**) is done on a continuous basis with exchanges, four times a day
- Continuous cyclic peritoneal dialysis (**CCPD**) is done during the night using a machine to make the exchanges while you sleep.

## Transplant

A kidney transplant is the preferred treatment method for kidney failure as it provides better long-term benefits for the patient. Unfortunately there is a severe shortage of donor organs and ADPKD patients may have to undergo dialysis before a suitable donor is available.

In transplantation, a healthy kidney is placed in the lower abdomen where it takes over the function of the failed kidneys. The diseased polycystic kidneys are normally left in place unless they are severely infected; removing them can be more complicated than the transplant and requires a separate operation.

The usual sources of kidneys are:

- Deceased donors
- Living-related donor, eg parent, child, sibling
- Living-unrelated donor, eg spouse, friend, good Samaritan

Living-related organs have the best outcomes, with some continuing to work well up to 20 years, thanks to modern immunosuppressive medications.

All donor organs, however, must be blood-type compatible.

**Note:** in ADPKD families, living-related donors are not accepted if less than 30 years, as there may be a risk of ADPKD in the donor.

## Managing pain

Pain is common in ADPKD patients and may be due to:

- **Acute** causes such as infection, bleeding from a cyst or kidney stones
- **Chronic** causes such as lower back pain or pressure pain from enlarging cysts.

Medication may include drugs containing paracetamol or aspirin (unless contra-indicated) – but not ibuprofen.

Non-pharmacological treatments may include posture exercises, heat or ice packs.

### **Avoid substances that are harmful to kidneys**

ADPKD patients should avoid using a class of drugs called NSAIDs (Non-Steroidal Anti-inflammatory Drugs), notably **Ibuprofen**. NSAIDs can accelerate loss of kidney function.

Decongestants should be avoided, as should Chinese and other herbal 'medicines' which may contain NSAIDs and other harmful substances.

### **Protect against flu**

Request a flu jab from the GP during the winter months which is recommended for people with ADPKD.

### **See a kidney specialist as early as possible and have regular check-ups**

According to the Department of Health, there is growing evidence about the **negative** effect of 'late referral' of patients with advanced impairment of kidney function.

Regular monitoring of kidney function is essential for the optimum management of ADPKD patients and helps avoid the problems associated with late referral – particularly because ADPKD is highly variable, symptoms are not always evident and laboratory tests are needed to detect complications. Therefore:

- Visit your GP regularly to have your blood pressure checked or obtain a BP monitor for home use
- Ensure you are regularly monitored by a renal consultant or nephrologist, for GFR measurement, BP control and dietary advice **at least annually**

### **What are the long-term cure prospects?**

Early treatments of complications, control of blood pressure, dialysis and kidney transplantation are the only methods of managing ADPKD.

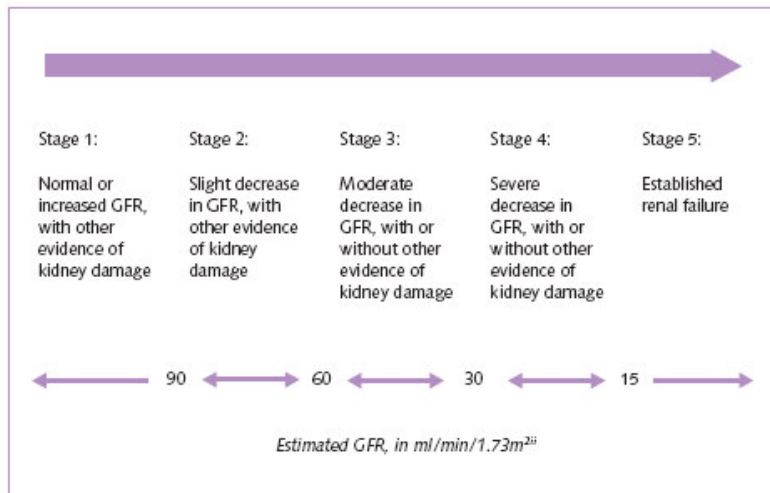
However, over the last 20 years there has been a considerable amount of worldwide research into the disease. More effective drugs are available for controlling high blood pressure and clinical trials are underway in the US on drugs that may reduce the growth of cysts.

## The Five Stages of Chronic Kidney Disease (CKD)

The stages of CKD are mainly based on measured or estimated GFR.

There are five stages but kidney function is normal in Stage 1 and minimally reduced in Stage 2.

The stages are described below:



Source: The Department of Health National Service Framework for Renal Services

## **Other Information and Help for ADPKD Patients**

### **The Polycystic Kidney Disease (PKD) Charity**

The PKD Charity funded the costs of production and printing of this booklet.

The charity is the only UK organisation dedicated to the support of people of PKD and their families; it also funds research into preventative treatments and a future cure.

For more information or to make a donation, contact:

The PKD Charity  
PO Box 141  
Bishop Auckland  
County Durham  
DL14 6ZD

[www.pkdcharity.org.uk](http://www.pkdcharity.org.uk)

Email: [info@pkdcharity.org.uk](mailto:info@pkdcharity.org.uk)

### **Other kidney charities and relevant associations**

National Kidney Federation UK

[www.kidney.org.uk](http://www.kidney.org.uk)

Tel: 0845 601 0209

Kidney Research UK

[www.nkrf.org.uk](http://www.nkrf.org.uk)

Tel: 0845 070 7601

The Blood Pressure Association UK

[www.bpassoc.org.uk](http://www.bpassoc.org.uk)

Genetic Interest Group UK

[www.gig.org.uk](http://www.gig.org.uk)

Tel: 020 7704 3141

The PKD Foundation USA

[www.pkdcure.org](http://www.pkdcure.org)

### **Acknowledgements**

The first edition of this booklet was written by Ms Kate Pettitt and Dr Anand K Saggarmalik in 1996 and further edited with the kind collaboration of Dr. Francesca Timio in 2002.

This issue was edited by Trustees and Medical Advisors of the PKD Charity.

The Charity would also like to thank Isabel Castellano who kindly drew some of the illustrations.