The commonest form of PKD is inherited
The commonest form of PKD is inherited

AUTOSOMAL DOMINANT POLYCYSTIC KIDNEY DISEASE

ADPKD
The commonest form of PKD is inherited

AUTOSOMAL DOMINANT POLYCYSTIC KIDNEY DISEASE

ADPKD

It is the commonest inherited kidney disease
POLYCYSTIC KIDNEY DISEASE

Some families with inherited kidney cysts... do not have ADPKD
POLYCYSTIC KIDNEY DISEASE

Some people with kidney cysts...

have not inherited them
AUTOSOMAL DOMINANT POLYCYSTIC KIDNEY DISEASE

What problems do the kidneys cause?

What other complications can there be?

How do we diagnose ADPKD?

What happens if the kidneys fail?
CYSTS IN ADPKD
The cysts are very small at birth
The cysts are very small at birth.

The kidney cysts are nearly always beginning to be visible on ultrasound scan by age 18 years.
The cysts are very small at birth.

The kidney cysts are nearly always beginning to be visible on ultrasound scan by age 18 years.

As the cysts increase in size and number they compress remaining kidney tissue.
The cysts are very small at birth

The kidney cysts are nearly always beginning to be visible on ultrasound scan by age 18 years

As the cysts increase in size and number they compress remaining kidney tissue

There are usually cysts in the liver as well
ADPKD
WHAT PROBLEMS DO THE KIDNEYS CAUSE?
ADPKD
WHAT PROBLEMS DO THE KIDNEYS CAUSE?

Some people get no symptoms at all
ADKD
WHAT PROBLEMS DO THE KIDNEYS CAUSE?

Haematuria

*Blood in the urine*
ADKD
WHAT PROBLEMS DO THE KIDNEYS CAUSE?

Haematuria

Bleed into cyst
Cyst infection
Rupture cyst
ADKD
WHAT PROBLEMS DO THE KIDNEYS CAUSE?

Haematuria

- Bleed into cyst
- Cyst infection
- Rupture cyst

Check for other causes of haematuria
COMPLICATIONS OF PKD

Urine infection
COMPLICATIONS OF PKD

Urine infection

Cyst infections may need long courses of antibiotics

Cyst aspiration if persistent infection

Long term antibiotics for frequent infections
ADPKD

WHAT PROBLEMS DO THE KIDNEYS CAUSE?

Stones
ADPKD
WHAT PROBLEMS DO THE KIDNEYS CAUSE?

Stones

- High fluid intake
- Lithotripsy
- Percutaneous stone removal
ADPKD
WHAT PROBLEMS DO THE KIDNEYS CAUSE?

Pain
ADPKD
WHAT PROBLEMS DO THE KIDNEYS CAUSE?

Pain

ACUTE
Bleeding, infection, stones
ADPKD

WHAT PROBLEMS DO THE KIDNEYS CAUSE?

Pain

ACUTE
Bleeding, infection, stones

CHRONIC
Mild or severe
ADPKD
WHAT PROBLEMS DO THE KIDNEYS CAUSE?

Pain

CHRONIC & SEVERE
ADPKD

WHAT PROBLEMS DO THE KIDNEYS CAUSE?

- Pain
  - Aspirate large cysts
    - Surgical cyst fenestration – laparoscopy
ADPKD

WHAT PROBLEMS DO THE KIDNEYS CAUSE?

Pain

Aspirate large cysts

Surgical cyst fenestration – laparoscopy

Surgical procedures do not affect decline in kidney function
ADPKD

WHAT PROBLEMS DO THE KIDNEYS CAUSE?

High blood pressure
ADPKD

WHAT PROBLEMS DO THE KIDNEYS CAUSE?

High blood pressure

BP goes up in early adult life

Treat the blood pressure

... to protect the kidneys

... to reduce cardiovascular risk
ADPKD
WHAT PROBLEMS DO THE KIDNEYS CAUSE?

High blood pressure

Treat the blood pressure
ADPKD
WHAT PROBLEMS DO THE KIDNEYS CAUSE?

High blood pressure

Treat the blood pressure

Lifestyle

Salt

Medicines
ADPKD

WHAT PROBLEMS DO THE KIDNEYs CAUSE?

Haematuria
Urine infection
Stones
Pain
High blood pressure
Kidney Failure
OTHER COMPLICATIONS OF ADPKD

LIVER CYSTS
OTHER COMPLICATIONS OF ADPKD

LIVER CYSTS

Multiple and vary in size

Do not cause liver failure
LIVER CYSTS IN ADPKD

Common problem is the size of the liver
LIVER CYSTS IN ADPKD

Common problem is the size of the liver

*Abdominal discomfort*

*Feeling full when you eat*

*Back ache*
LIVER CYSTS IN ADPKD

Big liver
LIVER CYSTS IN ADPKD

Big liver

Cyst infection
LIVER CYSTS IN ADPKD

Big liver

Cyst infection

Obstructive jaundice
LIVER CYSTS IN ADPKD

Reduce the size of the liver
LIVER CYSTS IN ADPKD

Reduce the size of the liver

Aspirate or fenestrate a single large cyst
LIVER CYSTS IN ADPKD

Reduce the size of the liver

Aspirate or fenestrate a single large cyst

Partial hepatectomy
10 years ago  3 years ago  Now
Liver resection & cyst fenestration
INTRACRANIAL ANEURYSMS IN ADPKD
INTRACRANIAL ANEURYSMS IN ADPKD

‘Berry’ aneurysms
Only in a few families (<10%)
INTRACRANIAL ANEURYSMS IN ADPKD

Only in a few families (<10%)

Often no symptoms
INTRACRANIAL ANEURYSMS IN ADPKD

Only in a few families (<10%)

More likely to bleed if blood pressure not controlled
INTRACRANIAL ANEURYSMS IN ADPKD

Only in a few families (<10%)

More likely to bleed if blood pressure not controlled

Most remain asymptomatic
INTRACRANIAL ANEURYSMS IN ADPKD

Only in a few families (<10%)

Often no symptoms

More likely to bleed if blood pressure not controlled

Most remain asymptomatic

May need surgery or ‘coil’
Who should be screened?
Who should be screened?
Who should be screened?

Only people with a family history of aneurysm
INTRACRANIAL ANEURYSMS IN ADPKD

Who should be screened?

Only people with a family history of aneurysm

Annual MR scan
Who should be screened?

Only people with a family history of aneurysm

Annual MR scan

If first scan negative...

Chance of a new aneurysm is about 3% at 10 yrs
Heart
Mitral valve prolapse in older age
not usually a problem
COMPLICATIONS OF ADPKD

Heart
Mitral valve prolapse in older age
not usually a problem

Gut
Diverticular disease
There are other inherited causes of cystic kidneys
DIAGNOSING ADPKD

There are other inherited causes of cystic kidneys

All less common than ADPKD
There are other causes of cystic kidneys

Simple cysts

Usually just a few cysts

Commoner with increasing age
HOW IS ADPKD DIAGNOSED?

Family history
HOW IS ADPKD DIAGNOSED?

- Family history
- Kidney failure
- Hypertension
- Sudden death
HOW IS PKD DIAGNOSED?

- Family history
- Scanning
SHOULD WE SCREEN FOR ADPKD?

Pros
• Knowledge concerning the diagnosis,
• Appropriate family planning,
• The ability to detect and treat complications associated with the disease,
• Reassurance of unaffected individuals
• Appropriate selection of unaffected relatives as possible donors for kidney transplantation.
SHOULD WE SCREEN FOR ADPKD?

**PRO**

- Knowing the diagnosis
- Family planning
- Treat complications early
- Reassure people who are not affected

Pros:
- Knowledge concerning the diagnosis,
- Appropriate family planning,
- The ability to detect and treat complications associated with the disease,
- Reassurance of unaffected individuals
- Appropriate selection of unaffected relatives as possible donors for kidney transplantation.
SHOULD WE SCREEN FOR ADPKD?

**PRO**
- Knowing the diagnosis
- Family planning
- Treat complications early
- Reassure people who are not affected

**CON**
- Effect on employment
- Life insurance
TESTING FOR ADPKD

ULTRASOUND SCAN
TESTING FOR ADPKD

ULTRASOUND SCAN

If ADPKD runs in your family, you have it if:

• 3 or more cysts - 15-39 yrs
• 2 or more cysts in each kidney - 40-59 yrs
• 4 or more cysts in each kidney – 60 yrs
TESTING FOR ADPKD

ULTRASOUND SCAN

If ADPKD runs in your family, you have it if:

- 3 or more cysts - 15-39 yrs
- 2 or more cysts in each kidney - 40-59 yrs
- 4 or more cysts in each kidney – 60 yrs

If ADPKD runs in your family, you have not got it if:

- Less than 2 cysts – age more than 40 yrs
HOW IS PKD DIAGNOSED?

Family history

Scanning

Genetic testing

There is still no ideal DNA test
WHAT PROBLEMS DOES ADPKD CAUSE FOR CHILDREN AND YOUNG ADULTS?

Often none at all
WHAT PROBLEMS DOES ADPKD CAUSE FOR CHILDREN AND YOUNG ADULTS?

Often none at all

Blood pressure begins to go up
WHAT PROBLEMS DOES ADPKD CAUSE FOR CHILDREN AND YOUNG ADULTS?

- Often none at all
- Blood pressure begins to go up
- Rarely - can be diagnosed in early childhood
ADPKD
WHAT PROBLEMS DO THE KIDNEYS CAUSE?

Kidney failure
ADPKD

WHAT PROBLEMS DO THE KIDNEYS CAUSE?

Kidney failure

It is unusual to get kidney failure before age 30 years
Kidney failure

Kidney failure is not inevitable
RISK OF PROGRESSION OF ADPKD
RISK OF PROGRESSION OF ADPKD

Male gender
RISK OF PROGRESSION OF ADPKD

- Male gender
- Hypertension

Pros
- Knowledge concerning the diagnosis,
- Appropriate family planning,
- The ability to detect and treat complications associated with the disease,
- Reassurance of unaffected individuals
- Appropriate selection of unaffected relatives as possible donors for kidney transplantation.
RISK OF PROGRESSION OF ADPKD

Male gender

Hypertension

Proteinuria
Pros
• knowledge concerning the diagnosis,
• appropriate family planning,
• the ability to detect and treat complications associated with the disease,
• reassurance of unaffected individuals
• appropriate selection of unaffected relatives as possible donors for kidney transplantation.

RISK OF PROGRESSION OF ADPKD

Male gender
Hypertension
Proteinuria
Increasing renal size
RISK OF PROGRESSION OF ADPKD

- Male gender
- Hypertension
- Proteinuria
- Increasing renal size
- Genotype (PKD 1 versus PKD 2)
Declining eGFR over time.
Declining kidney function
He is admitted to hospital with severe loin pain and visible haematuria.
The graph illustrates the decline in eGFR over years, with key events marked:

- Clot obstruction
- Obstruction relieved
- Renal replacement therapy

The data points show a steady decline in eGFR from 1986 to 1994, followed by interventions at 1994 and 1996 that halt the decline.
APDKD: WHAT HAPPENS WHEN THE KIDNEYS FAIL?

Renal replacement therapy

DIALYSIS

KDINEY TRANSPLANT
For many people it is preferable to have independent treatment in the community.
For many people it is preferable to have independent treatment in the community.

Kidney Transplant
For many people it is preferable to have independent treatment in the community.

- Kidney Transplant
- Home Haemodialysis
- Peritoneal Dialysis
For many people it is preferable to have independent treatment in the community.

- Kidney Transplant
- Home Haemodialysis
- Peritoneal Dialysis
- Conservative Kidney Care
RENAL REPLACEMENT THERAPY

For many people it is preferable to have independent treatment in the community:

- Kidney Transplant
- Home Haemodialysis
- Peritoneal Dialysis
- Conservative Kidney Care

For some:

Haemodialysis in a hospital or satellite unit
RENAL REPLACEMENT THERAPY

For many people it is preferable to have independent treatment in the community

- Kidney Transplant
- Home Haemodialysis
- Peritoneal Dialysis
- Conservative Kidney Care

For some:

- Haemodialysis in a hospital or satellite unit

Patient Choice
There is a shortage of deceased donor organs
We are doing more living donor transplants

But there is still a shortage of living donor kidneys
DIALYSIS IN APDKD

CAPD may not be possible
NEPHRECTOMY IN ADPKD
NEPHRECTOMY IN ADPKD

Not routine -

EPO

Fluid balance

Major surgery
NEPHRECTOMY IN ADPKD

Only nephrectomy if major problem with…..

Pain
Infection
Bleeding
Size