# 

#### **PRE-PREGNANCY**

Pre-implantation Genetic Testing (PGT) is available to those with a known family history of Cystic Renal Disease.

#### PREGNANCY

Scans during pregnancy may reveal signs of PKD. Genomic testing may be discussed and offered via amniocentesis.

#### **BABIES**

New babies may be offered genomic testing if signs of PKD were noticed during antenatal scans or newborn checks.

#### **CHILDREN**

Children attending paediatric nephrology clinic may be offered genetic testing.

#### **YOUNG ADULTS**

Teens and young adults at risk of PKD should be made aware of testing when transitioning to adult services.

### **FAMILY HISTORY**

When an individual has confirmed PKD, a genetic diagnosis can be helpful for family members who may also be affected.

#### **NEW SYMPTOMS**

New onset of PKD symptoms is typically diagnosed by ultrasound scan and family history but may be confirmed by genomic testing.



## POLYCYSTIC **KIDNEY** DISEASE

There are many opportunities for genomic testing for PKD throughout a person's lifetime

GENOMIC

TESTING

KNOWLEDGE

PKD is an inherited disorder passed from parents to children. In the case of the most common form, ADPKD, if one parent passes the affected gene to a child they have a 50% chance of getting the disease.

Knowing if you're affected is important as most problems, such as high blood pressure, pain and UTIs, can be treated with medication. The medication Tolvaptan can be used to slow down the formation of cysts and protect kidney function.

When kidney function deteriorates further, treatment options include dialysis and kidney transplantation.

"Having the knowledge helped

to change our viewpoint and

understand our next steps for

testing our other child"







SUPPORT

#### **HOW TO DECIDE**

To help you make an informed decision about the pros and cons of genetic testing, and decide if it is right for you, visit the PKD Charity website:



#### WHO TO SPEAK TO

If you are under the care of a kidney healthcare professional please speak to them about testing. If you think you could be affected please speak to your GP.

Patient information poster developed by NHS East Genomics September 2023 V1

"Our son had an ultrasound ... and a genetic test in which no cysts were dentified. His genetic test was negative for PKD [which meant] we knew he didn't need to be monitored in the same way as his brothers and dad"

"Not knowing either way was affecting my mental health...I was anxious. We need better mental health support for young people affected by PKD, even if you do not have the condition"

> **People's** experience of genetic testing ...