

Kidney Health Information

Autosomal Recessive Polycystic Kidney Disease (ARPKD)

This condition is also known as Infantile Polycystic Kidney Disease. It is completely different from the adult form known as Autosomal Dominant Polycystic Kidney Disease ([ADPKD](#)).

What is ARPKD?

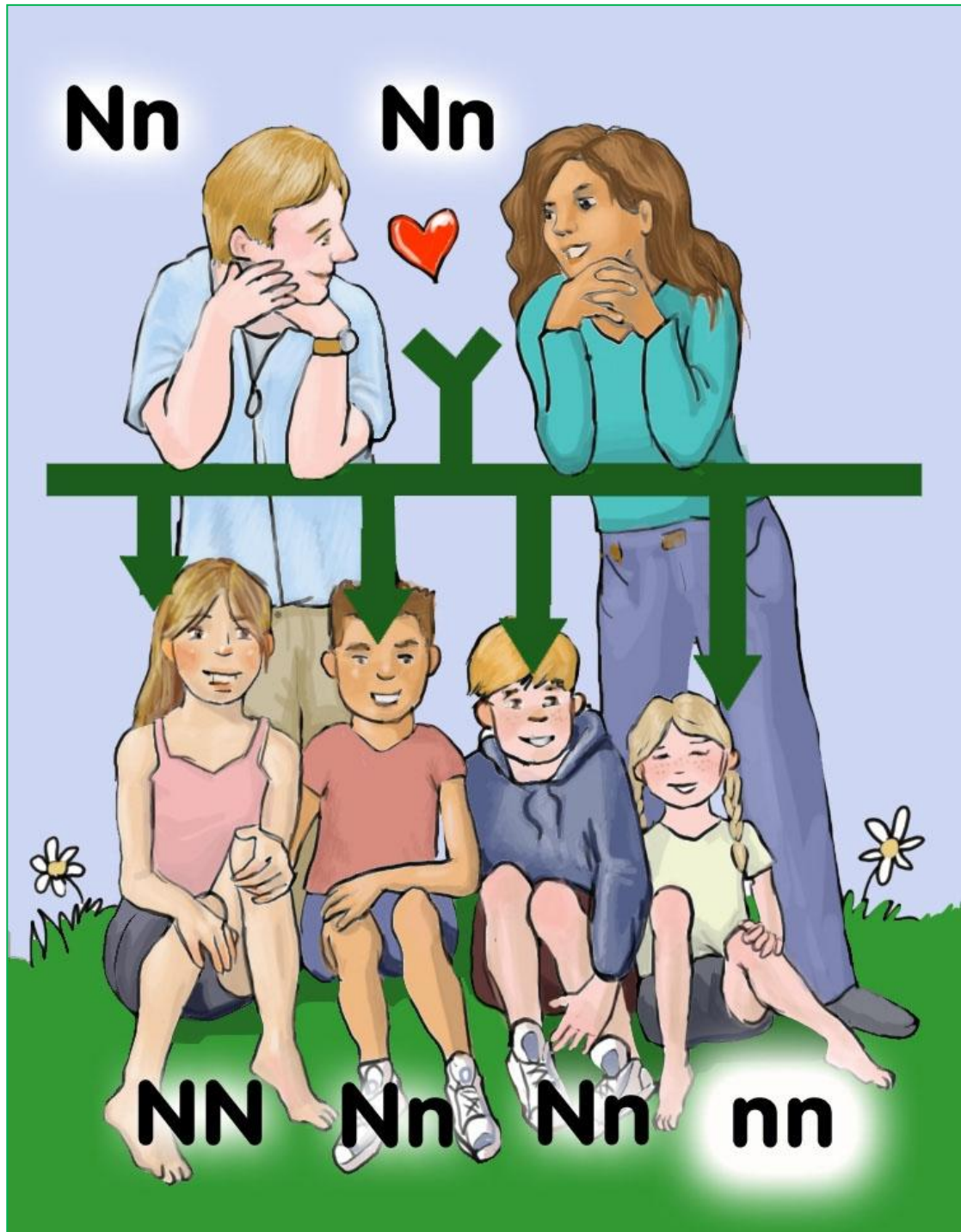
A rare inherited disease in which the kidneys are damaged by cysts in both kidneys. Small cysts form inside the collecting tubules, which are inside the nephron. The cysts enlarge, forming balloon-like swellings. The more nephrons are affected, the greater the loss of kidney function.

How is it caused?

ARPKD is a rare genetic condition caused by an abnormality in the Fibrocystin gene.

- A 'recessive disorder', the baby inherits a copy of the affected gene from each 'carrier' parent. As the baby has inherited two recessive genes, it is affected
- The parents are unaffected, as they each carry just one copy of the affected gene. So both parents will not have ARPKD

- The chances of any other child being born with ARPKD if their parents have already had one with ARPKD are one in four



In the UK, between one in 10,000 to one in 40,000 babies are affected. Once the baby survives the first few weeks of life, the outlook is good. At one year old, the baby has a 70% chance of not needing dialysis or a transplant by 15 years of age. For the other 30%, dialysis or a transplant may be needed by ten years of age. Later on, the age at which kidney failure develops varies greatly between patients.

Other Problems

- The liver is affected as well and liver disease may be severe
- Scarring may lead to blockage of veins in the liver, which enlarges
- This is known as congenital hepatic fibrosis
- Sometimes other kidney conditions are present with congenital hepatic fibrosis
- Severely affected babies may have underdeveloped lungs, due to the size of the kidneys during pregnancy

Pre-natal diagnosis

Many cases of ARPKD are found in pregnancy, by routine ultra sound scanning or at birth. For those families already affected by ARPKD pre-natal diagnosis may be available.

The first signs of ARPKD vary greatly – at its most severe, the baby is affected during pregnancy and the kidneys become greatly enlarged. The enlargement can be so large that the babies breathing can be impeded after birth. As a result up to about half of babies with ARPKD die at birth or very soon after.

Finding out More

[PKD Charity](#) Tel 01388 665004

[National Kidney Federation](#) Helpline 01909 544999

[Transplant Support Network](#) Tel 0800 027 4490/1 (two lines)

[Contact a Family](#) Tel 0808 808 3555

[Eric](#) – Education and Resources for improving Childhood Continence
Tel: 0845 370 8008

Some research we have supported

2006 Dr Mark Pfhul University of Leicester

[Towards a better understanding of a protein important in polycystic kidney disease by structural investigation of its domain organisation.](#)

**2003 Prof Judith Goodship Institute for Human Genetics,
University of Newcastle**

[The role of inversin in primary cilia and renal cyst formation](#)

2002 Prof A Woolf & Miliyun Chiu, ICH, UCL London

[Roles of galectin-3 in Autosomal Recessive Polycystic Disease \(ARPKD\) and Acute Tubular Necrosis.](#)

Please be aware that we have made every effort to ensure this information is accurate, however we cannot guarantee that there are no mistakes. Also, the best management plans for individual patients may vary from those outlined here. Only the doctors caring for the patient will be able to advise on this. Please consult your own doctor.

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