PKD1 & Autosomal Dominant Polycystic Kidney Disease

What is Autosomal Dominant Polycystic Kidney Disease?

- Autosomal Dominant Polycystic Kidney Disease (ADPKD) is an inherited disease caused by changes in one of two PKD genes – PKD1 and PKD2.
- This change in the PKD gene causes your kidneys to become enlarged and dysfunctional.
- Signs and symptoms usually begin in adulthood and include high blood pressure, urinary tract infections, blood and/or protein in the urine, pain in your sides (flank pain) and abdominal pain.

What is PKD1, and how do changes in PKD1 affect the kidneys?

- While the exact function of the PKD1 is not known, it is believed to play a role in cell growth and promoting normal kidney development and function.
- When there is a change in PKD1, there is an abnormal growth of cells in the kidney. This causes numerous fluid-filled sacs called cysts to form.
- These cysts continue to grow and eventually interfere with the functions of the kidney, causing the signs and symptoms of ADPKD mentioned above.
- As the disease progresses it can result in kidney failure and you may require dialysis or a kidney transplant – on average this occurs at around 55 years of age.

Do these changes have effects on other parts of the body?

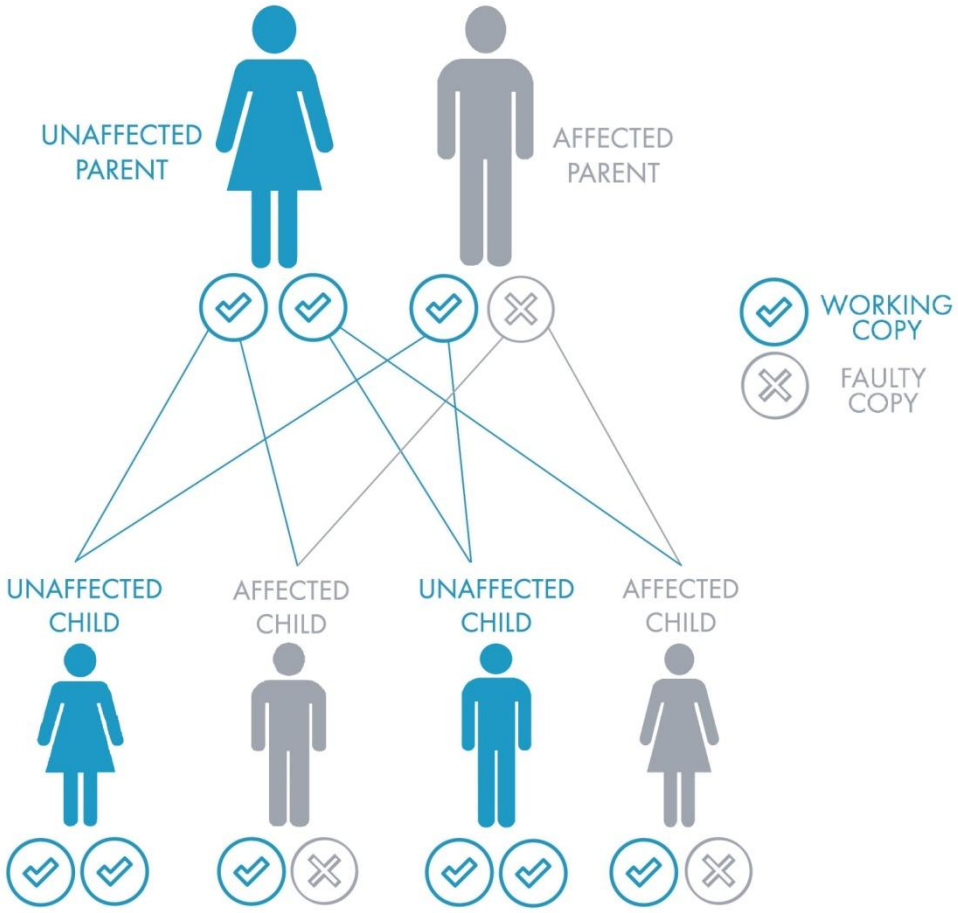
- Cysts may also form in other organs including the liver, pancreas, thyroid gland and spleen.
- Hernias, in which areas of the belly bulge out, are fairly common and usually occur just above the belly button or in the groin area.
- While incredibly rare, the most serious possible complication is a bulging blood vessel in the brain (aneurysm). If left untreated, it can have serious consequences, so your doctor will recommend getting a scan for this.

How is ADPKD treated?

- Treatment involves managing symptoms and slowing down the progression of the disease.
- This includes, but is not limited to:
  - Treating high blood pressure through lifestyle changes, diet changes and medicines.
  - Treating kidney infections with antibiotics.
  - Pain killers.
- Surgery may be required in the case of hernia or aneurysm.

How is this change passed down through a family?

- You have two copies of PKD1 – one copy from each of your parents.
- To have ADPKD you must inherit at least one faulty copy of the PKD1 gene from an affected parent.
- Each child of an affected parent has a 1 in 2 (50%) chance of inheriting the disease.
- There is usually a family history of ADPKD, but in about 10-15% of cases there is no known cases of the disease in the family. This may be because affected family members are not aware they have the disease or were not diagnosed in their lifetime.



Should my family members be tested?

- Adult family members should talk to their doctor about getting tested, and it is recommended that they have a discussion with a genetic counselor before any genetic testing is carried out.
- It is not generally recommended that children are tested when they do not have symptoms. However, you may be advised to have your children's blood pressure checked by their doctor or nurse once a year.