
HNF1 Beta & Renal Cysts and Diabetes Syndrome

What is Renal Cysts and Diabetes Syndrome?

- Renal Cysts and Diabetes Syndrome is an inherited condition associated with changes in the gene HNF1B.
- It is associated with impaired development of several organs including the kidneys and the development of a form of diabetes which begins during your teens or in early adulthood.

What is HNF1 Beta, and how do changes in HNF1 Beta affect the kidneys?

- HNF1 Beta is a gene that is involved in the development of the pancreas, kidney, liver, urinary tract and reproductive system.
- When this gene changes, it can result in structural abnormalities and impaired function of these organs.
- The extent of dysfunction is incredibly variable, even among affected members of the same family.
- Fluid-filled sacs called cysts form in the kidneys, which can interfere with kidney cells ability to filter the blood. Over time this can lead to kidney failure, though this only occurs in about 12% of cases.
- Kidneys may be under-developed, or one kidney may be missing. In very rare cases, the kidneys do not develop at all – affected babies are unlikely to survive outside of the womb.

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

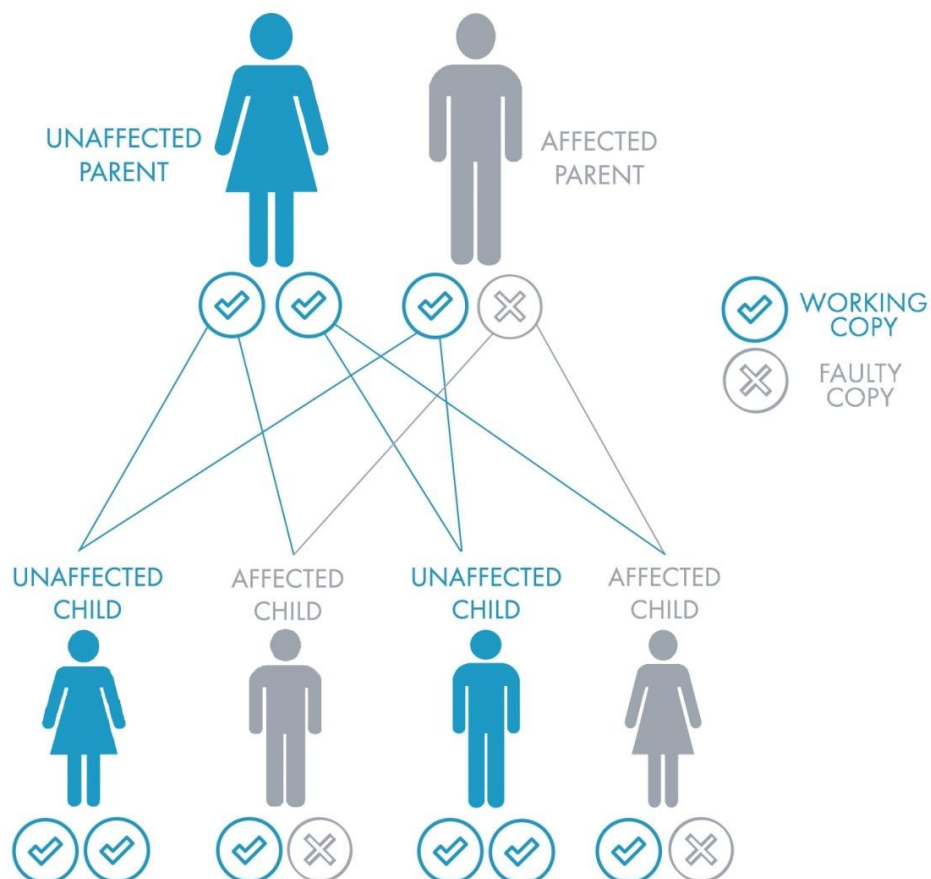
- The pancreas is often affected. Cells which are involved in the regulation of blood sugar levels become dysfunctional, resulting in diabetes. However, not everyone with changes in HNF1 Beta develops diabetes.
- The pancreas can also waste away (atrophy). This is a problem, as the pancreas plays an important role in the digestion of food through the production of enzymes. When it is smaller than normal, not enough enzymes are produced, and this results in loose stools and unintentional weight loss.
- The uterus can be an abnormal shape or there may even be two uteruses present. It is possible for woman with this kind of abnormality to have a successful pregnancy.
- The liver is also affected, but this does not generally result in significant liver disease.
- Due to the effects on kidney function, there can be a build-up of a substance called uric acid. This deposits on the joints and causes gout. This causes severe pain in the joints.

How is Renal Cysts and Diabetes Syndrome treated?

- If the kidneys fail, dialysis or kidney transplant is required.
- If diabetes develops, a controlled diet, insulin injections and careful monitoring are important parts of management.
- Insufficient levels of pancreatic enzymes can be replaced through taking tablets at meal times.
- Gout is treated with drug therapies such as allopurinol.

How is this change passed down through a family?

- You have two copies of HNF1 Beta – one copy from each of your parents.
- To develop this syndrome you must inherit at least one faulty copy of the HNF1 Beta gene from an affected parent.
- Each child of an affected parent has a 1 in 2 (50%) chance of inheriting the disease.
- However, most cases of this condition are due to spontaneous gene changes in people with no family history of the condition.



Should my family members be tested?

- Family members may be advised to undergo genetic testing.
- It is recommended that this is discussed with a genetic counsellor before testing is carried out.