AVP & Familial Diabetes Insipidus

What is Familial Diabetes Insipidus?

- Familial Diabetes Insipidus (FDI) is an inherited condition which causes an imbalance in water intake and excretion.
- This leads to the production of excessive amounts of urine and consequently, dehydration and excessive thirst.
- Affected individuals need to urinate frequently, which can disrupt daily activities and sleep.
- Although excessive urination is also a feature, this is not to be confused with diabetes mellitus, which is a very different condition.

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

- The AVP gene provides instructions for the production of anti-diuretic hormone (ADH), which controls the balance between fluid intake and urine excretion by the kidney.
- When you are adequately hydrated, the body lowers production of this hormone as you don't need to reabsorb more water. Levels increase again when you become less hydrated.
- When this gene is changed, there are lower levels of this hormone available. This means that even when you need water, the kidney will continue to excrete any fluids you consume as from its perspective, low levels of ADH mean the body is already hydrated.

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

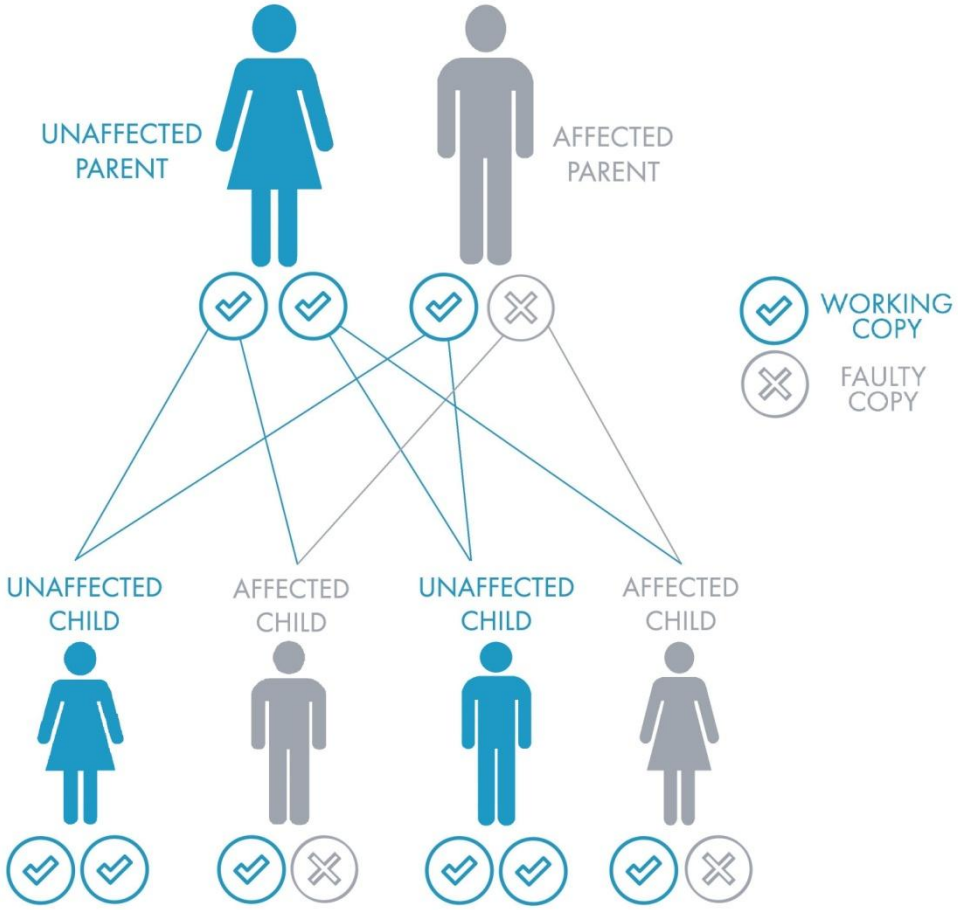
- People with FDI can become dehydrated very quickly. This can lead to constipation and dry skin.
- If untreated, severe dehydration can result in confusion, low blood pressure, seizures and coma.

How is FDI treated?

- The most common treatment for diabetes insipidus is a drug called desmopressin, which increases ADH levels.
    - It is important that use of this drug is monitored as it can lead to excessive water retention and dangerously low salt levels in the blood, which can potentially cause brain injury.
- You may be encouraged to follow a low salt low protein diet.

How is this change passed down through a family?

- You have two copies of AVP – one copy from each of your parents.
- To develop FDI you must inherit at least one faulty copy of the AVP gene from an affected parent.
- Each child of an affected parent has a 1 in 2 (50%) chance of inheriting the disease.



Should my family members be tested?

- If a family history is identified, it may be advised for family members to undergo genetic testing.
- Before this testing is carried out, it is recommended that family members have a discussion with a genetic counsellor.