BBS10 & Bardet-Biedl Syndrome

What is Bardet-Biedl syndrome?

- Bardet-Biedl syndrome is an inherited disorder caused by changes in the BBS genes, resulting in a wide range of effects throughout the body.
- This can include childhood obesity, extra toes or fingers, vision impairment, intellectual disability and kidney abnormalities

What is BBS10, and how do changes in BBS10 affect the kidneys?

- The BBS10 gene is involved in the production of cilia, tiny finger-like projections that are found on the surface of many cells. They are involved in cell movement and the perception of sensations (e.g. sight, hearing, smell).
- When BBS10 changes, the cilia of kidney cells do not work properly, and the kidney is not able to reabsorb water properly.
- Over time, kidney function decreases. However, kidney failure only occurs in a small minority of affected individuals, usually in later life.
- Early symptoms include increased urination and thirst. Later, there is the formation of fluid-filled sacs called cysts in the kidney, and protein is present in the urine.

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

- A number of effects are associated with this syndrome; however, symptoms vary from person to person.
  - Loss of vision is a major feature of the syndrome. Issues with night vision begin by mid-childhood, followed by the development of blind spots in the peripheral vision. Most become blind by their teen years or early adulthood.
  - Obesity tends to develop around 2 – 3 years of age.
  - Extra toes or fingers may be present.
  - Genitals can be under-developed or poorly functioning. This may be associated with infertility.
  - Children with this syndrome often have learning disorders and may have speech and language problems.
- Other features can include diabetes, high blood pressure, heart defects, bowel disease, neurological problems affecting coordination and movement, dental abnormalities and behavioural disorders.

How is Bardet-Biedl syndrome treated?

- There is no cure for this syndrome. Treatment focuses on managing symptoms.
  - There is no way to prevent gradual vision loss; however, vision aids, mobility training and planning for blindness early can improve quality of life.
  - Obesity may be managed through education, diet and exercise from an early age.
Early intervention, special education and speech therapy may be required to manage intellectual disability.
Hormone levels should be monitored when puberty starts in order to see whether replacement therapy is required.
Surgery for the extra toes/fingers or genital abnormalities may be needed.
Kidney transplants can be successful; however, the drugs given after the transplant may contribute to obesity.

How is this change passed down through a family?

- You have two copies of BBS10.
- To develop Bardet-Biedl syndrome, two faulty copies of the BBS10 gene must be inherited, one from each parent – they are “carriers” of the faulty gene and do not have the disease themselves.
- Each child of carrier parents has a 1 in 4 (25%) chance of inheriting the disease.
- If a child receives only one copy of a faulty gene, they themselves become carriers. They will not have Bardet-Biedl syndrome but may pass on that faulty gene to their own offspring.
- A small number of cases seem to require an additional mutation on a separate gene to develop the syndrome.

Should my family members be tested?

- Genetic testing may be made available to family members.
- It is recommended to have a discussion with a genetic counsellor prior to testing.