
BBS1 & 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 BBS1, and how do changes in BBS1 affect the kidneys?

- The BBS1 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 BBS1 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.

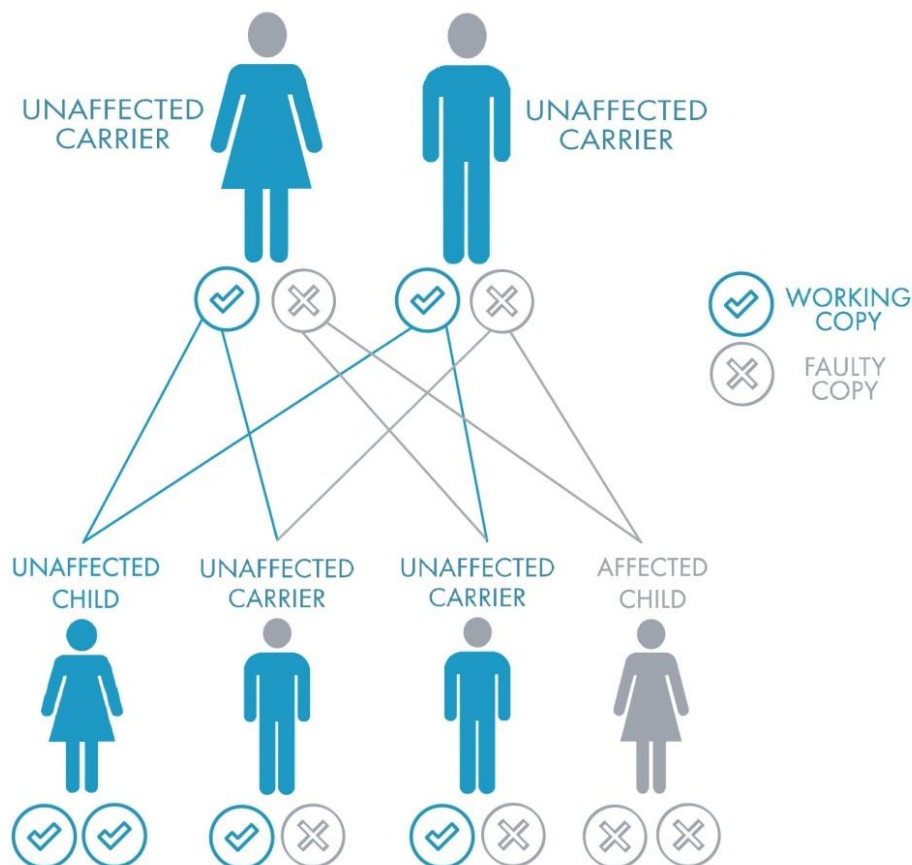
How is Bardet-Biedl syndrome treated?

- 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 BBS1.
- To develop Bardet-Biedl syndrome, two faulty copies of the BBS1 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.