Bardet-Biedl syndrome

What is Bardet-Biedl syndrome?

Bardet-Biedl syndrome (BBS) is a syndromic inherited retinal disease (IRD) that affects several parts of the body. BBS is also known as Laurence-Moon-Bardet-Biedl syndrome.

It is a rare syndrome, thought to affect between 1 in 140,000 and 1 in 160,000 newborns in North America and Europe, and less than 50 new cases reported each year in the USA.[1] There are over 25 genes that are known to cause BBS, with the most common being BBS1 (23%), followed by BBS10 (15%) and BBS2 (10%).[2]

How is it identified?

BBS is diagnosed when a person with retinitis pigmentosa (RP) also shows systemic symptoms, as outlined below.

What are the symptoms?

The diagnosis of BBS is often confirmed when visual symptoms start, which is generally in early childhood.

People with BBS develop a type of RP which initially causes poor night vision, and then leads to loss of peripheral vision, tunnel vision, and eventual loss of central vision. As with other forms of RP, this vision loss can be variable, with some people losing nearly all sight, and others maintaining their vision for longer.

In addition to the vision loss, people with BBS can show:

- Obesity (particularly in the trunk of the body)
- Polydactyly (extra toes or fingers)
- Webbing between fingers and toes
- Developmental delays, such as mild intellectual impairment and/or delayed emotional development, can be seen in around half of people with BBS
- Kidney disease
- Abnormalities of the genitalia

Due to the high chance of obesity, people with BBS are also prone to health conditions associated with weight gain, such as Type 2 diabetes, high blood pressure and high cholesterol.

What is the cause of Bardet-Biedl syndrome and how is it inherited?

Bardet-Biedl syndrome can result from mutations in at least 18 different genes in the body. These are commonly called *BBS* genes. All of these genes are involved in producing hair-like structures called cilia within cells, which allow the cells to move and detect sensory input.

The inheritance of Bardet-Biedl syndrome is autosomal recessive. Autosomal recessive inheritance means each parent is a carrier, but is not affected by the disease themselves. Each of their children will have a 25% chance of being affected, with males and females having an equal chance of being affected. The children have a 50% chance of being a carrier (having one healthy and one mutated copy of the gene).

What treatments are available?

At this stage, there is no cure for BBS, therefore treatment focusses on managing symptoms. For example:

- Vision impairments – Low vision aids and mobility training are beneficial.
- Weight management – A dietician can work with the individual and/or their family to create an appropriate diet regimen.
- Kidney problems – There are several types of kidney diseases associated with BBS and the treatment varies accordingly. Kidney issues can be treated with medication and in rare instances, surgery (including, if needed, kidney transplantation).
- Polydactyly – Extra fingers and toes, or webbing between the digits, are usually removed surgically in childhood.

A number of emerging treatments are being developed that may assist people with BBS in the future.

References
(1) Bardet-Biedl syndrome. Medline Plus. Accessed 3 January 2024. medlineplus.gov/genetics/condition/bardet-biedl-syndrome
(2) Weihbrecht K. Bardet-Biedl syndrome. In: Genetics and Genomics of Eye Disease. Academic Press; 2020:117-136.

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Contact Retina Australia
info@retinaaustralia.com.au
(03) 9650 5088