RETINA australia

Bardet-Biedl syndrome (BBS)

Bardet-Biedl syndrome (BBS) is a condition that affects several parts of the body. It runs in families and may severely impair the sufferer. BBS is also known as Laurence-Moon-Bardet-Biedl syndrome.

What are the Symptoms of Bardet-Biedl Syndrome?

One of the major features of Bardet-Biedl syndrome is loss of vision. Loss of vision occurs as the retina (the light sensing tissue or layer at the back of the eye) gradually fails. The problems appear initially as difficulty in seeing in the dark. This is seen in childhood as a slow onset condition. This is followed by development of blind spots in vision that leads to falls and knocks initially.

Over time, these blind spots enlarge and turn into a tunnel where only the centre is visible. With time the central clear area also clouds. By the time the child reaches adolescence or early adulthood, he may become legally blind. Another characteristic feature can be weight problems. There is typical abdominal weight gain that begins in early childhood and continues throughout life. This may give rise to other complications like type 2 diabetes, high blood pressure and abnormally high levels of cholesterol.

Typically, there may also be presence of extra fingers and/or toes along with intellectual disability, learning challenges, and abnormalities of the reproductive organs or external genitalia.

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

Bardet-Biedl syndrome is a genetically inherited condition. It results from mutations in at least 14 different genes in the body. These are commonly called BBS genes. It is found that these genes help make the hair like cellular structures called cilia. These cilia are involved in cell movement, different chemical signalling pathways, perception of sensory input (such as sight, hearing, and smell) etc.

The BBS genes code for proteins that maintain the functions of the cilia. Defects in these genes lead to defective cilia. The inheritance of Bardet-Biedl syndrome is autosomal recessive. This means parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene but have no symptoms of the condition (carriers 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 aides and mobility training are beneficial.
- Weight – A dietician can work with the family to create an appropriate diet regimen.

- Speech disorders – Speech therapy can help children with their communication skills and parents can be taught exercises to use at home to improve their child's ability to communicate.
- Kidney problems – There are several types of kidney problems associated with BBS and the treatment varies accordingly. Kidney issues can be treated with medication and in rare instances, surgery (transplantation).
- Polydactyly – Skin tags can be tied off at birth and extra digits can be removed surgically.

We acknowledge www.news-medical.net as the source of information shown above.

The Marshfield Clinic in Wisconsin, U.S.A. has established the Clinical Registry Investigating Bardet-Biedl Syndrome.
Enrolment into the Registry can be undertaken through http://www.bbs-registry.org/.