Bardet-Biedl Syndrome
A guide for patients

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This booklet has been produced by the Euro-WABB project. The Euro-WABB project initiated in 2011 following an international workshop organised by Association Syndrome de Wolfram. The project aims to support efficient diagnosis, treatment, and research for Wolfram, Alström, Bardet-Biedl (WABB) and other rare syndromes. The purpose of this leaflet is to provide information and guidance on Wolfram Syndrome to Wolfram Syndrome patients and their families. We would like to sincerely thank all of the partners, clinicians and families who have contributed to this document.
Bardet-Biedl Syndrome (BBS) is a genetic disorder that can cause a set of conditions including obesity, diabetes, retinal dystrophy, polydactyly, and renal failure. Some patients also have developmental delays and learning difficulties.

**Why is it called Bardet-Biedl Syndrome?**

A syndrome is the name given to a condition where features occur in a consistent pattern, and where the cause is not understood. Bardet-Biedl Syndrome (BBS) is named after Georges Louis Bardet, a French physician (born 1885) and after Artur Biedl, a Hungarian pathologist and endocrinologist (born 1869). Since the original descriptions, over 200 patients have been described in the world medical literature.

**How common is it?**

Bardet-Biedl Syndrome affects about 1 in 160,000 people, so it is very rare. A specialist physician may only see one affected child in a professional lifetime.

**What causes Bardet-Biedl Syndrome?**

Bardet-Biedl Syndrome is an inherited genetic condition caused by a gene mutation. Many genes have been identified as being affected in Bardet-Biedl Syndrome. The syndrome is classed as a “ciliopathy” because the gene mutations affect a protein in the body which works the cilia structure of cells. Originally, doctors thought that Bardet-Biedl was caused by recessive genes, although it has now been demonstrated that sometimes more than one mutation in more than one gene can cause Bardet-Biedl Syndrome. So for a child to be affected, he/she may inherit sometimes several abnormal copies of one or more BBS genes.
What are the features of Bardet-Biedl Syndrome?

Bardet-Biedl Syndrome is very varied and presents differently for different people. The following are some of the most common features:

**Polydactyly**

Polydactyly refers to when a child is born with extra fingers or toes. Syndactyly can also occur in Bardet-Biedl Syndrome and this refers to when digits such as fingers and toes are partially fused together.

**Obesity**

Some children with Bardet-Biedl Syndrome can develop early-onset obesity. This is part of the condition and can be controlled with healthy diets and exercise. Obesity can lead to complications later in life such as fatty liver disease later on in childhood.

**Retinal Dystrophy**

Retinal dystrophy refers to when the retina at the back of the eyes breaks down over time and there are abnormalities with the rods and cones in the eye. This can cause nystagmus or “wobbly eyes”. The visual impairments associated with Bardet-Biedl Syndrome are progressive conditions which can lead to impaired night-vision, tunnel vision and patients often experience early onset blindness.

**Hypogonadism**

Hypogonadism refers to underdeveloped and functional genitals in males and females.

**Renal Problems**

Renal abnormalities can occur in patients with Bardet-Biedl Syndrome. This can include cystic kidneys which are detected antentatally or in early childhood. The kidneys can have other irregular shapes and functionally, patients report excessive thirst and urine production. Cystic kidneys which are. These difficulties may lead to chronic renal failure, which needs treatment.
Some children with Bardet-Biedl Syndrome have developmental delay and may have physical difficulties for instance delays in learning to walk or difficulties with coordination. Also some children have mild to moderate learning difficulties and may need extra support at school. Bardet-Biedl Syndrome can also cause speech delays and deficits in speech.

**Other Features**

There are a variety of other conditions which may also affect people with Bardet-Biedl Syndrome. Diabetes Mellitus can occur. This is when the body cannot convert glucose (sugar) to energy, because the pancreas is not making enough of a hormone called Insulin. Glucose therefore stays in the blood or is passed out in urine and is not used up by the body. Symptoms of Diabetes Mellitus include thirst, frequent passing of urine, and weight loss.

Conductive hearing loss which is sometimes known as “glue ear” is also a possible symptom of Bardet-Biedl Syndrome.

Dental abnormalities, heart conditions and liver problems are all also possible symptoms of Bardet-Biedl Syndrome.

**Diagnosing Bardet-Biedl Syndrome**

As Bardet-Biedl Syndrome is so rare not all doctors will be aware of it and because the condition is so complex and variable for different individuals it can be difficult to recognise. Early diagnosis and management of conditions leads to better health outcomes for patients and improved quality of life. Therefore it is important that you show this information sheet to your doctors so they know what information you have. For further clinical information for you or your doctor, the Euro-WABB project has published a clinical management guideline for Bardet-Biedl Syndrome which is available on the Euro-WABB website (www.euro-wabb.org).

Most of the features of Bardet-Biedl Syndrome will exhibit during childhood. If retinal dystrophy and obesity are both present during childhood, before the age of 15 then an individual may have Bardet-Biedl Syndrome.
The current treatment for Bardet-Biedl Syndrome involves treating the various presenting symptoms separately:

* Polydactyly - extra digits are usually surgically removed when a child is very young.
* Obesity can be treated with dietary advice from a dietician and through regular exercise.
* Unfortunately there is no cure for retinal dystrophy. Early diagnosis can allow the complications to be managed better.
* Hypogonadism can be treated with hormone replacement therapy to replace the hormones that the body does not naturally make.
* Renal problems can be treated depending on the type of anomaly a person has. There is also treatment for chronic kidney disease and renal failure if this develops.
* Learning difficulties can be managed effectively if they are recognised early. Speech difficulties can be improved via a Speech Therapist and extra help at school might be required.
* Diabetes Mellitus can be controlled with insulin injections. These replace the insulin in the body and allow glucose to be converted to energy for the cells to use.

Once a child has been diagnosed with Bardet-Biedl Syndrome regular health appointments and assessments will be required. These assessments are to manage the presenting symptoms of the syndrome and also to test for the related symptoms that we know about so that they can be diagnosed and managed as early as possible.

Yearly eyesight and hearing tests are recommended for Bardet-Biedl patients to track the progress of retinal dystrophy and any hearing loss. Blood test and urine tests can monitor any renal problems. For obesity and diabetes, diet and exercise advice will be given along with blood tests to look at average blood glucose levels over a few weeks and other treatments to manage any complications of diabetes. Cognitive assessments for learning difficulties and regular testing for any issues arising from the heart or liver are also recommended.
Unfortunately at the moment there is no cure for Bardet-Biedl Syndrome. The current research that is taking place is to understand why mistakes in the Bardet-Biedl genes cause the syndrome. There are research groups in America, France, Germany and the UK all investigating this problem.

The chances of parents having another child affected by Bardet-Biedl Syndrome are difficult to predict. This is because doctors originally thought it was a recessive disorder. It has now been demonstrated that some forms of Bardet-Biedl Syndrome require more than one mutation in more than one gene locus. So for a child to be affected, he/she has to inherit sometimes several abnormal copies of one or more BBS genes. It is possible to test if an unborn child is affected during pregnancy.

For further information on Bardet-Biedl Syndrome please see:
- **Lawrence-Moon-Bardet-Biedl society** ([www.lmbbs.org.uk](http://www.lmbbs.org.uk))
  The Society supports families and communicates with health professionals involved in their care.
- **Ciliopathy Alliance** ([www.ciliopathyalliance.org](http://www.ciliopathyalliance.org))
  Information and support website for diseases caused by defects in the function or structure of cilia.
  This article on Gene reviews provides a good review of the Syndrome. The article is written by Professor Phil Beales, a leading expert in Bardet-Biedl Syndrome.
- **Orphanet** ([www.orpha.net](http://www.orpha.net))
  Orphanet is an online database of rare diseases and related services provided through Europe. It contains information on over 5 000 conditions and lists specialised clinics, diagnostic tests, patient and organizations, research projects and clinical

For any queries relating to the information supplied in this guide please contact Euro-WABB at [euro-wabb@bch.nhs.uk](mailto:euro-wabb@bch.nhs.uk)
For further clinical information for you or your doctor please see the Wolfram Clinical Management Guidelines published on the Euro-WABB website.
There are a number of patient support groups and clinicians around Europe who are interested in Bardet-Biedl Syndrome. This contact sheet provides information on groups and clinicians that have been involved in the Euro-WABB project.

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<th>Country</th>
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For all other countries or for information on genetic diagnostic laboratories please contact [euro-wabb@bch.nhs.uk](mailto:euro-wabb@bch.nhs.uk)