

### What is Bardet-Biedl syndrome?

Bardet-Biedl syndrome is an inherited disease characterized by progressive vision loss, obesity, birth defects, learning disabilities, and behavioral problems. The symptoms associated with Bardet-Biedl syndrome are due to the abnormal functioning of cilia, which are hair-like structures found at the surface of many cells.<sup>3</sup> BBS is also known as Laurence-Moon-Bardet-Biedl syndrome.<sup>1</sup>

### What are the symptoms of Bardet-Biedl syndrome and what treatment is available?

The signs and symptoms of Bardet-Biedl syndrome are variable, even within families, and may include:<sup>1,2</sup>

- Progressive vision loss (childhood onset leading to legal blindness by adolescence or early adulthood)
- Polydactyly (extra fingers/toes)
- Obesity
- Learning disabilities
- Behavioral problems
- Genitourinary malformations
- Kidney abnormalities
- Hypertension
- Diabetes
- Congenital heart defects
- Complete or partial loss of sense of smell

There is no cure for Bardet-Biedl syndrome. Treatment includes visual aids and education programs for the visually impaired, early intervention and special education for learning disabilities, dietary therapy, and exercise programs for obesity, as well as surgical correction for birth defects.<sup>2</sup>

### How is Bardet-Biedl syndrome inherited?

Bardet-Biedl syndrome can be caused by mutations in one of at least 19 different genes, including *BBS1*, *BBS2*, and *BBS10*, which are responsible for more than 50% of reported cases.<sup>2</sup> Bardet-Biedl syndrome is primarily an autosomal recessive disease; fewer than 10% of cases result from a complex pattern of inheritance which requires three or more mutations in two different Bardet-Biedl syndrome genes.<sup>2</sup>

Based on an autosomal recessive pattern of inheritance, an individual who inherits one mutation in a BBS gene is a carrier and is typically not expected to have related health problems. An individual who inherits two mutations in the same BBS gene, one mutation from each parent, is expected to be affected with Bardet-Biedl syndrome.

Based on an autosomal recessive pattern of inheritance, if both members of a couple are carriers of mutations in the same gene, the risk for an affected child is 25% in each pregnancy; therefore, it is especially important that the reproductive partner of a carrier be offered testing.

### Who is at risk for Bardet-Biedl syndrome?

BBS can occur in individuals of all races and ethnicities. It is more common in the Bedouin population of Kuwait and on the island of Newfoundland. Bardet-Biedl syndrome is estimated to affect approximately 1 in 140,000 individuals worldwide<sup>1</sup>; the risk of being a carrier of a *BBS1* mutation is calculated to be 1 in 390, and the risk of being a carrier of a *BBS10* mutation is calculated to be 1 in 418.<sup>2</sup> In the Ashkenazi Jewish population, the risk of being a carrier of *BBS2* is 1 in 136.<sup>4</sup>

Having a relative who is a carrier or who is affected can increase an individual's risk of being a carrier. Consultation with a genetics health professional may be helpful in determining carrier risk and appropriate testing.

### What does a positive test result mean?

If a gene mutation is identified, an individual should speak to a physician or genetics health professional about the implications of the result and appropriate testing for the reproductive partner and at-risk family members.

### What does a negative test result mean?

A negative result reduces, but does not eliminate, the possibility that an individual carries a gene mutation. The likelihood of being a carrier is also influenced by family history, medical symptoms, and other relevant test results.

There are at least 19 genes associated with BBS. Inheritest tests for mutations in the most common BBS-related gene(s), but it does not assess a person's risk for being a carrier of mutations in the other, less common BBS-related genes.

### Where can I get more information?

Bardet Biedl Syndrome Family Association: <http://www.mlmorris.com/lmbbs/index.html>

National Organization for Rare Disorders (NORD): <https://rarediseases.org/rare-diseases/bardet-biedl-syndrome/>

### References

1. Bardet-Biedl syndrome. *Genetics Home Reference*. Available at <http://ghr.nlm.nih.gov/condition/bardet-biedl-syndrome>. Accessed January 13, 2016.
2. Forsythe E and Beales PL. Bardet-Biedl Syndrome. *Gene Reviews*. Available at <http://www.ncbi.nlm.nih.gov/books/NBK1363/>. Accessed January 13, 2016.
3. Baker K, Beales PL. 2009. Making sense of cilia in disease: The human ciliopathies. *Am J Med Genet Part C Semin Med Genet* 151C:281–295.
4. Fedick A *et al*. Carrier frequency of two *BBS2* mutations in the Ashkenazi population. *Clinical Genetics*. 2014; 85(6):578-582