BARDET-BIEDL SYNDROME

GENETIC TESTING

LabCorp
Laboratory Corporation of America

What is Bardet-Biedl syndrome?

Bardet-Biedl syndrome (BBS) is an inherited disease characterized by progressive vision loss, obesity, birth defects, learning disabilities, and behavioral problems. The symptoms associated with BBS are likely due to the abnormal functioning of cilia, which are hair-like structures found on the surface of many cells of the body. BBS is also known as Laurence-Moon-Bardet-Biedl syndrome.[1]

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

The signs and symptoms of Bardet-Biedl are variable, even within families, and may include[1,2]:

- Progressive vision loss (childhood onset leading to legal blindness by adolescence or early adulthood)
- Polydactyly (extra fingers/toes)
- Obesity
- Learning disabilities
- Behavioral problems
- Genital anomalies
- Kidney abnormalities
- Hypertension
- Diabetes
- Congenital heart defects (structural abnormalities of the heart present at birth)
- Complete or partial inability to smell

There is no cure for BBS. 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.[2]

How is Bardet-Biedl syndrome inherited?

BBS can result from mutations in (at least) 14 different genes, including *BBS1* and *BBS10*, which are responsible for more than 40% of reported cases.[1] BBS is primarily an autosomal recessive disease with fewer than 10% of cases following an atypical pattern of inheritance.[2]

In autosomal recessive 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 BBS.

In the autosomal recessive form of BBS, 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, but it is more common in the Bedouin population of Kuwait and on the island of Newfoundland. In the general North American population, BBS is estimated to affect approximately 1 in 140,000 individuals[1]; however, 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.

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 14 genes associated with BBS. Carrier screening 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?

- Laurence-Moon-Bardet-Biedl Family Network: **www.mlmorris.com/lmbbs/index.html**
- National Organization for Rare Disorders: **www.rarediseases.org/rare-disease-information/rare-diseases/byID/988/viewAbstract**

Note: This material is provided for general information purposes only. It is not intended as a substitute for medical advice and/or consultation with a physician or technical expert.

References

1. Bardet-Biedl syndrome. Genetics Home Reference Web site. Available at: http://ghr.nlm.nih.gov/condition/bardet-biedl-syndrome. Accessed March 1, 2012.
2. Waters AM and Beales PL. Bardet-Biedl Syndrome. Gene Reviews Web site. Available at http://www.ncbi.nlm.nih.gov/books/NBK1363/. Accessed March 1, 2012.

LabCorp
Laboratory Corporation of America

www.LabCorp.com

Contact Us

For more information about LabCorp, the testing services we provide, and where to find a specimen collection lab near you, visit **www.labcorp.com.**