

What is Bardet-Biedl syndrome?

Bardet-Biedl syndrome is a complex disorder that affects many parts of the body including the retina. Individuals with this syndrome have a retinal degeneration similar to retinitis pigmentosa (RP).

What are the symptoms?

The diagnosis of Bardet-Biedl syndrome is often confirmed in childhood when visual problems due to RP are discovered. The first symptom of RP is night blindness. Night blindness makes it difficult to see in low light levels. RP then causes a progressive loss of peripheral (side) vision. Peripheral vision loss is often referred to as tunnel vision. Individuals with Bardet-Biedl also experience central vision loss during childhood or adolescence. RP symptoms progress rapidly and usually lead to severe visual impairment by early adulthood.

In addition to RP, polydactyly (extra fingers and/or toes) and obesity are defining characteristics of Bardet-Biedl syndrome. The condition is usually first suspected when a child is born with polydactyly. Subsequent RP symptoms and obesity confirm the diagnosis. Extra fingers and toes are

usually removed in infancy or early childhood. Slight webbing (extra skin) between fingers and between toes is also common. Most individuals have short, broad feet as well. Obesity may be present by childhood and is usually limited to the trunk of the body. Many individuals are also shorter than average.

Approximately half of all individuals with Bardet-Biedl syndrome experience developmental disabilities ranging from mild impairment or delayed emotional development to mental retardation. The degree of mental retardation can range from mild cognitive disability to severe mental retardation. Individuals may also experience renal (kidney) disease. Renal abnormalities can affect the structure and the function of the kidneys and can lead to severe renal impairment.

Upon reaching adulthood, males with Bardet-Biedl syndrome can have small genitalia. Females with Bardet-Biedl can experience irregular menstrual cycles.

Is it an inherited disease?

Bardet-Biedl syndrome is genetically passed through families by the autosomal recessive pattern of

Bardet-Biedl Syndrome

inheritance. In this type of inheritance both parents, called carriers, have one gene for the syndrome paired with one normal gene. Each of their children then has a 25 percent chance (or 1 chance in 4) of inheriting the two Bardet-Biedl genes (one from each parent) needed to cause the disorder. Carriers are unaffected because they have only one copy of the gene.

What testing is available?

Genetic testing is available for BBS. It helps assess the risk of passing the disorder from parent to offspring. It also helps with attaining an accurate diagnosis. A patient with an accurate diagnosis is in a better position to keep track of new findings, research developments, and treatment approaches. A genetic diagnosis can also help a person qualify for clinical trials. More information on genetic testing is available at www.FightBlindness.org. Researchers, including those funded by the Foundation, have identified 18 genes that are linked to Bardet-Biedl syndrome, giving them targets for treatment development.

What treatment is available?

Some emerging treatments for RP that are designed to work independent from the disease-causing gene mutations may benefit people with BBS. These may include certain drugs, gene

therapies, and stem-cell treatments. For the latest research advances for RP which may apply to people with BBS, refer to the Foundation publication *Retinitis Pigmentosa: Research Advances*.

To manage the complications of renal disease associated with Bardet-Biedl syndrome, every individual with the disorder should be examined by a nephrologist, a physician who specializes in kidney diseases.

Are there other related syndromes?

Bardet-Biedl syndrome is often confused with Laurence-Moon syndrome. Individuals with Laurence-Moon syndrome almost always experience neurologic problems but rarely polydactyly. Polydactyly is a defining feature of Bardet-Biedl syndrome, while neurologic problems almost never occur. Laurence-Moon syndrome is extremely rare; only a few cases have been documented. Because of the similarity of these syndromes, Bardet-Biedl syndrome is often referred to as Laurence-Moon/Bardet-Biedl syndrome or Laurence-Moon/Biedl syndrome.