Bardet-Biedl Syndrome

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Bardet-Biedl syndrome is a disorder that affects many parts of the body. The signs and symptoms of this condition vary among affected individuals, even among members of the same family.

Keywords: genetic conditions

1. Introduction

Vision loss is one of the major features of Bardet-Biedl syndrome. Loss of vision occurs as the light-sensing tissue at the back of the eye (the retina) gradually deteriorates. Problems with night vision become apparent by mid-childhood, followed by blind spots that develop in the side (peripheral) vision. Over time, these blind spots enlarge and merge to produce tunnel vision. Most people with Bardet-Biedl syndrome also develop blurred central vision (poor visual acuity) and become legally blind by adolescence or early adulthood.

Obesity is another characteristic feature of Bardet-Biedl syndrome. Abnormal weight gain typically begins in early childhood and continues to be an issue throughout life. Complications of obesity can include type 2 diabetes, high blood pressure (hypertension), and abnormally high cholesterol levels (hypercholesterolemia).

Other major signs and symptoms of Bardet-Biedl syndrome include the presence of extra fingers or toes (polydactyly), intellectual disability or learning problems, and abnormalities of the genitalia. Most affected males produce reduced amounts of sex hormones (hypogonadism), and they are usually unable to father biological children (infertile). Many people with Bardet-Biedl syndrome also have kidney abnormalities, which can be serious or life-threatening.

Additional features of Bardet-Biedl syndrome can include impaired speech, delayed development of motor skills such as standing and walking, behavioral problems such as emotional immaturity and inappropriate outbursts, and clumsiness or poor coordination. Distinctive facial features, dental abnormalities, unusually short or fused fingers or toes, and a partial or complete loss of the sense of smell (anosmia) have also been reported in some people with Bardet-Biedl syndrome. Additionally, this condition can affect the heart, liver, and digestive system.

2. Frequency

In most of North America and Europe, Bardet-Biedl syndrome has a prevalence of 1 in 140,000 to 1 in 160,000 newborns. The condition is more common on the island of Newfoundland (off the east coast of Canada), where it affects an estimated 1 in 17,000 newborns. It also occurs more frequently in the Bedouin population of Kuwait, affecting about 1 in 13,500 newborns.

3. Causes

Bardet-Biedl syndrome can result from mutations in at least 14 different genes (often called BBS genes). These genes are known or suspected to play critical roles in cell structures called cilia. Cilia are microscopic, finger-like projections that stick out from the surface of many types of cells. They are involved in cell movement and many different chemical signaling pathways. Cilia are also necessary for the perception of sensory input (such as sight, hearing, and smell). The proteins produced from BBS genes are involved in the maintenance and function of cilia.

Mutations in BBS genes lead to problems with the structure and function of cilia. Defects in these cell structures probably disrupt important chemical signaling pathways during development and lead to abnormalities of sensory perception. Researchers believe that defective cilia are responsible for most of the features of Bardet-Biedl syndrome.

About one-quarter of all cases of Bardet-Biedl syndrome result from mutations in the *BBS1* gene. Another 20 percent of cases are caused by mutations in the *BBS10* gene. The other BBS genes each account for only a small percentage of all cases of this condition. In about 25 percent of people with Bardet-Biedl syndrome, the cause of the disorder is unknown.

In affected individuals who have mutations in one of the BBS genes, mutations in additional genes may be involved in causing or modifying the course of the disorder. Studies suggest that these modifying genes may be known BBS genes or other genes. The additional genetic changes could help explain the variability in the signs and symptoms of Bardet-Biedl syndrome. However, this phenomenon appears to be uncommon, and it has not been found consistently in scientific studies.

3.1. The genes associated with Bardet-Biedl syndrome

- BBS1
- BBS10
- CEP290
- MKKS

3.2. Additional Information from NCBI Gene

- ARL6
- BBS12
- BBS2
- BBS4
- BBS5
- BBS7
- BBS9
- MKS1
- TRIM32
- TTC8

4. Inheritance

Bardet-Biedl syndrome is typically inherited in an autosomal recessive pattern, which means both copies of a BBS gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

5. Other Names for This Condition

- BBS

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