WHAT YOU SHOULD KNOW ABOUT BARDET-BIEDL SYNDROME

WHAT IS BARDET-BIEDL SYNDROME (BBS)?

Often diagnosed in childhood or adolescence, BBS is an inherited disease causing progressive loss of night and peripheral vision from retinitis pigmentosa (RP).

The retina is a thin piece of tissue lining the back of the eye.

Electrical signals are transferred to the brain via the optic nerve.

Rod and cone photoreceptors in the retina convert light into electrical signals.

HOW IS BBS INHERITED?

Bardet-Biedl syndrome is autosomal recessive. That means it is genetically passed when both parents have one gene for the syndrome paired with one normal gene.

There is a 25% chance of their child inheriting the two BBS genes. 18 BBS causing genes have been identified.

GENETIC TESTING

Genetic testing helps with attaining an accurate diagnosis. It also helps assess the risk of passing the disorder to offspring. A patient with an accurate diagnosis is in a better position to understand which emerging treatment approaches and clinical trials are most appropriate for them.

WHAT TO EXPECT WITH BARDET-BIEDL SYNDROME:

The diagnosis of Bardet-Biedl syndrome is often made at birth or in childhood when problems due to retinitis pigmentosa or other problems related to the syndrome are discovered.

OTHER RELATED SYMPTOMS:

Symptoms vary from person to person.

VISION RELATED SYMPTOMS:

Night Blindness
Loss of Peripheral Vision
Central Vision Loss

Polydactyly
Extra fingers and toes are usually removed in infancy or early childhood.

Developmental Disabilities
Mild impairment, delayed emotional development, or intellectual disability.

Obesity
May be present by childhood and is usually limited to the trunk of the body.

Renal (Kidney) Disease
Renal abnormalities can affect the structure and the function of the kidneys.

To learn more about living with Bardet-Biedl syndrome, finding a specialist, genetic testing or clinical trials, visit FightingBlindness.org or call 888-332-3667.