WHAT YOU SHOULD KNOW ABOUT

RETINITIS PIGMENTOSA

WHAT IS RETINITIS PIGMENTOSA?

Retinitis pigmentosa (RP) refers to a group of inherited retinal diseases, often leading to legal and sometimes complete blindness. Forms of RP and related diseases include Usher syndrome, Leber congenital amaurosis, and Bardet-Biedl syndrome, among others. An estimated 100,000 people in the U.S. have RP.

The retina is a thin piece of tissue lining the back of the eye. Rod and cone photoreceptors in the retina convert light into electrical signals that the brain interprets as vision.

WHAT TO EXPECT WITH RETINITIS PIGMENTOSA:

Symptoms depend on whether rods or cones are initially involved:

Rods: In most forms of RP, rods are affected first, causing loss of peripheral vision and loss of night vision. Vision becomes more constricted over time.

Cones: If and when the disease progresses and cones become affected, visual acuity, color perception, and central vision are diminished.

HOW IS RETINITIS PIGMENTOSA INHERITED?

Genetic mutations can be passed from parent to offspring through one of three genetic inheritance patterns:

1. Autosomal Recessive: Both parents carry one copy of the mutated gene and one normal copy, but have no symptoms themselves (unaffected carriers).

2. Autosomal Dominant: Only one parent is affected with one mutated copy of the gene. A child has a 50% chance of being affected through the inheritance of the mutated gene.

3. X-linked: The mutated gene for the disease is located on the X chromosome. Female carriers have a 50% chance of passing the disease gene to their daughters, who become carriers, and a 50% chance of passing the gene to their sons, who are then affected by the disease.

Affected males will never pass an X-linked disease to their sons. However, they will always pass their affected chromosomes to their daughters who will become carriers.

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