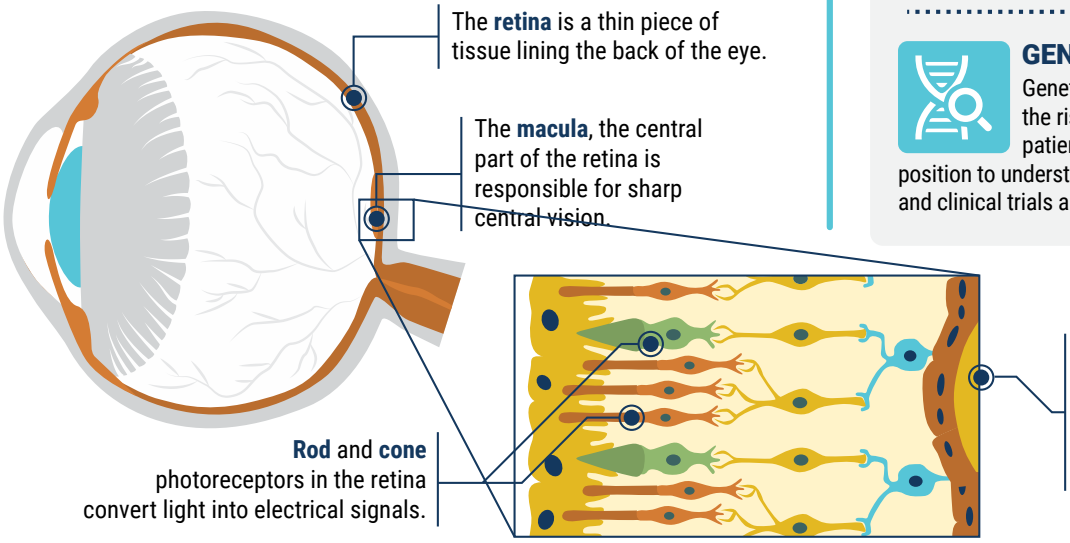
WHAT YOU SHOULD KNOW ABOUT

BEST DISEASE

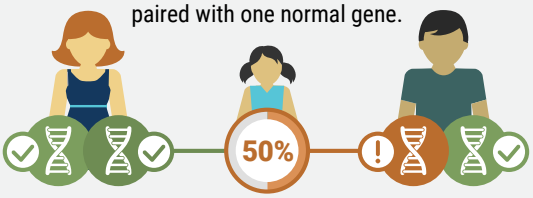
WHAT IS BEST DISEASE?

Best disease, also known as vitelliform macular dystrophy, is an inherited retinal disease causing macular degeneration, characterized by loss of central vision, as well as the ability to perceive colors and details.



HOW IS BEST DISEASE INHERITED?

Best disease is usually autosomal dominant. That means it is **genetically passed** when one parent has one disease causing gene, BEST1 (VMD2), paired with one normal gene.



There is a **50% chance** that the affected parent will pass the disease-causing gene to their child. The unaffected partner will only pass normal genes.

GENETIC TESTING

Genetic testing helps confirm the diagnosis and the risk of passing the condition 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 BEST DISEASE:

THINGS TO NOTE:

- Usually diagnosed during childhood.
- Severity of loss varies from person to person.
- Vision often deteriorates to about 20/100 later in life.
- Can be detected by retina specialists.
- Cause accumulation of yellow flecks in the retina.
- Does not always affect both eyes equally.

STAGES OF BEST DISEASE:

A **cyst forms** beneath the macula, like a sunny-side-up egg. Visual acuity may remain normal or near for many years.

The cyst can eventually **rupture**. Fluid and yellow deposits spread throughout the macula, having a scrambled egg appearance.

The **macula** and the RPE begin to atrophy (degenerate) causing further vision loss.

FOUNDATION FIGHTING BLINDNESS

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