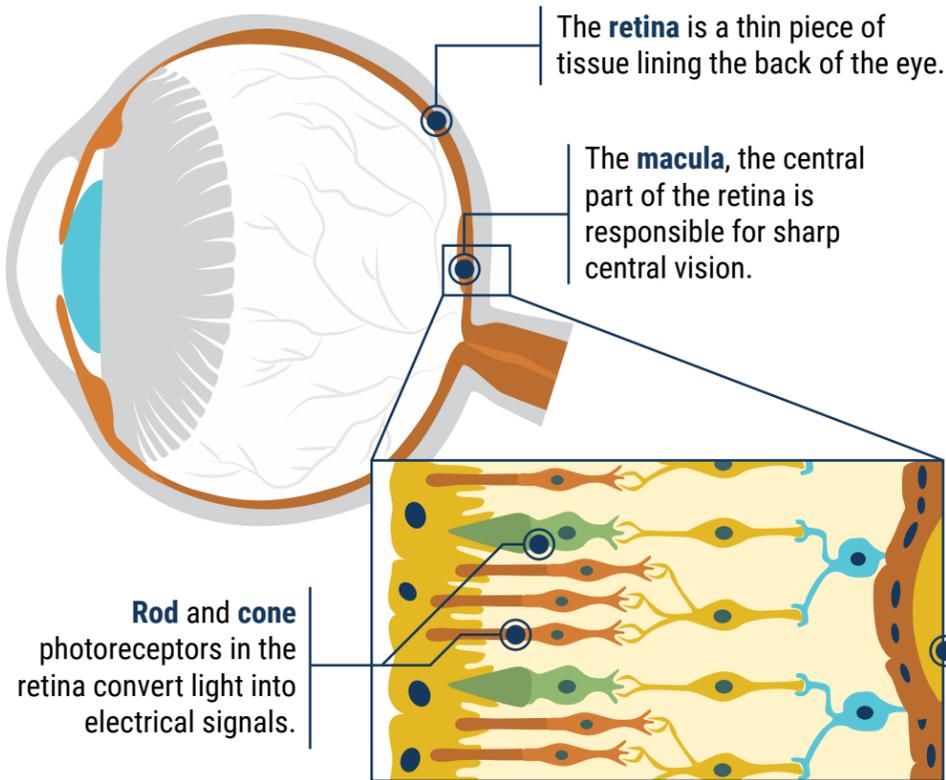


# BEST DISEASE

## 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.

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### 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.

In the initial stages of Best disease, a **yellow cyst** forms under the **retinal pigment epithelium (RPE)** beneath the macula. The RPE is a layer of cells that provides support for photoreceptors.

## WHAT TO EXPECT WITH BEST DISEASE:

### STAGES OF BEST DISEASE:

- 1** A **cyst** forms beneath the macula, like a sunny-side-up egg. Visual acuity may remain normal or near for many years.
- 2** The cyst can eventually **rupture**. Fluid and yellow deposits spread throughout the macula, having a scrambled egg appearance.
- 3** The **macula** and the **RPE** begin to atrophy (degenerate) causing further vision loss.

### What someone with Best Disease may see.

- Loss of central vision
- Reduced ability to perceive colors
- Reduced ability to perceive details

### THINGS TO NOTE:

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