

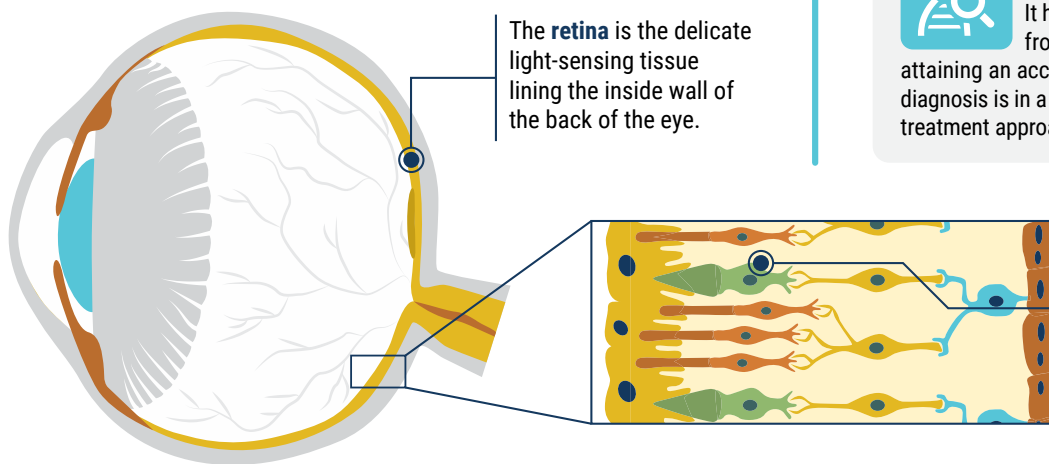
# WHAT YOU SHOULD KNOW ABOUT

# ACHROMATOPSIA

## WHAT IS ACHROMATOPSIA?

Achromatopsia is an inherited **retinal condition** affecting **cone photoreceptors**, the cells in the retina that provide vision in lighted conditions, central and color vision, and the ability to perceive details (e.g., read, recognize faces).

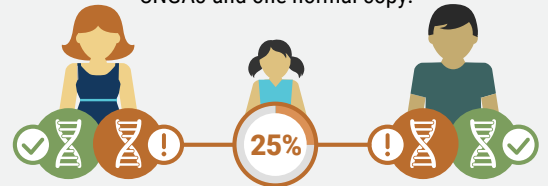
Achromatopsia is not progressive and is usually diagnosed in early childhood.



**Cones** provide vision in lighted conditions, central and color vision, and the ability to perceive details.

## HOW IS ACHROMATOPSIA INHERITED?

Achromatopsia is inherited when both parents have one **mutated copy of a specific gene** such as CNGB3 or CNGA3 and one normal copy.



The child has a **25% chance** of inheriting the two copies of the mutated gene, (from each parent) causing the disease.



### GENETIC TESTING

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

## WHAT TO EXPECT WITH ACHROMATOPSIA:



**Extreme Light Sensitivity**



**Reduced Visual Acuity**



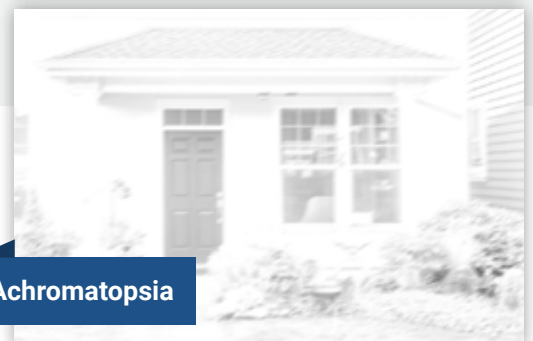
**Color Discrimination**

### LIGHT-FILTERING GLASSES

People with the condition wear glasses with tinted lenses to filter out the type of light that is uncomfortable. Different patients have different light-filtering needs.



Normal Vision



Achromatopsia