Achromatopsia in Children

This material will help you understand achromatopsia and how it is treated.

**What is achromatopsia?**

Achromatopsia is a visual disorder that causes decreased vision. Other symptoms of achromatopsia include:

- Sensitivity to light
- No color vision
- Nystagmus ("shaking" or "jiggling" of the eyes)

It is sometimes called “day blindness” because it is easier for people with achromatopsia to see in dim light.

**How does achromatopsia affect my child's vision?**

The degree of low vision varies among children. Your child may have moderate vision loss (20/120 to 20/80) or be legally blind (20/200 or less). However, achromatopsia does not cause total blindness. The condition is also not progressive. This means that it will not get worse over time.

**What causes achromatopsia?**

Achromatopsia is a genetic condition that affects the retina. The retina is the “film” of the eye. It captures images to send to the brain, making the picture your child sees. The retina is made up of cells called rods and cones. In achromatopsia, your child's cones do not work properly.

Scientists have found four genes that are involved in achromatopsia. Even if the parents do not have the condition, they can carry a gene for it. If the child inherits the gene from both parents, then he/she will have achromatopsia.
How is achromatopsia diagnosed?
Achromatopsia can be diagnosed by an eye doctor. The eye doctor will examine your child's retina and color vision. If your child's eye doctor suspects he/she have achromatopsia, the diagnosis can be confirmed by a special test called electroretinography (ERG).

How is achromatopsia treated?
There is no cure for achromatopsia. Research is looking into gene therapy as a possibly treatment in the future. If your child has a refractive error (like nearsightedness or farsightedness), glasses can help improve your child's vision. However, glasses will not cure the achromatopsia. Red lenses can help reduce sensitivity to light. Low vision aids like magnifying glasses and telescopes are also available.

How will achromatopsia affect my child in school?
In school, your child may find it hard to read from the blackboard. This can make learning more difficult. However, most children with achromatopsia can be successful in normal classrooms with only small changes like seating location. Your child may find it easier to sit in the front of the class and away from windows. Using large print books and magnifying glasses may also help. Talk to your child’s school about what kind of resources and services are available. Even if your child is not yet in school and has low vision, there are early intervention programs available through public schools.