



Achromatopsia

WHAT IS ACHROMATOPSIA?

Achromatopsia is a non-progressive and hereditary visual disorder which is characterized by the absence of color vision, decreased vision, light sensitivity, and nystagmus. The cause of this disorder is absence of functioning cones (photoreceptors) in the retina. Patients with complete achromatopsia are only able to perceive black, white and gray shades. Their world consists of different shades of gray ranging from black to white, rather like only seeing the world as black and white. Patients with incomplete achromatopsia may perceive some limited colors.

WHAT ARE THE TYPES OF ACHROMATOPSIA?

Achromatopsia has two types; the complete type in which there is no functional cones in the retina at all. These patients will have severe visual symptoms. The incomplete type in which there is some functional cones and patients will have less severe visual symptoms than the complete type.

WHAT VISION DOES A CHILD WITH ACHROMATOPSIA HAVE?

Children with achromatopsia will have reduced vision (typically 20/200 or less), no color vision (they perceive only black, white and gray shades), sensitivity to light (photophobia) and the presence of nystagmus (shaking of the eyes). Achromatopsia is sometimes called 'Day Blindness', as these children see better in subdued light. Children with incomplete Achromatopsia may have better vision (typically 20/120 to 20/80) and usually have less severe visual symptoms. Moreover, children may show paradoxical constriction of the pupil in the dark, usually have high hyperopic refractive errors (need for plus power glasses). Nystagmus and photophobia, marked in infancy, may improve with age. Achromatopsia is a non-progressive disorder, which means the vision is usually stable over time.

HOW COMMON IS ACHROMATOPSIA?

This condition affects approximately one in 30,000 live births. Its prevalence varies in different parts of the world. Because there is a genetic link, it is more common in regions where there is a high rate of consanguineous marriages (marriages between relatives) and in the eastern Pacific islands of Pingelap (see the book, 'The Island of the Colour blind' by Dr Oliver Sacks).



ARE RED-GREEN COLOR BLINDNESS (COLOR DEFICIENCY) AND ACHROMATOPSIA THE SAME CONDITION?

No, red-green color deficiency patients have difficulty distinguishing red and green colors. People with red-green color deficiency have otherwise normal visual acuity and do not have complete lack of color perception.

WHAT CAUSES ACHROMATOPSIA?

Achromatopsia is a genetic disorder in which a child is born with nonfunctioning cones. The cones are special photoreceptor cells in the retina that absorb different color lights. There are three types of cones that are responsible for normal color vision. These are the red cones, the green cones, and the blue cones. A balanced distribution of these cells is necessary for normal color vision. A child born with non-functioning cones will have achromatopsia. Mutations in the following genes are known to cause achromatopsia: ATF6, CNGA3, CNGB3, GNAT2, PDE6C and PDE6H.

HOW IS THE DIAGNOSIS MADE?

The diagnosis will be made by your ophthalmologist. Initially, family history and symptoms such as light sensitivity and reduced vision will provide clues essential to the diagnosis. The retinal examination may be normal in appearance. The color vision tests commonly performed in the clinic are the Ishihara pseudoisochromatic tests, H-R-R tests, Farnsworth panel D15 color test and the City University tests. Additional testing such as optical coherence tomography (OCT), fundus autofluorescence, visual fields, as well as electroretinogram (ERG) to measure cone function are also important in making the diagnosis for this condition. Genetic testing can be performed to test for the mutations in the most common genes to confirm the diagnosis.

WHAT ARE THE CHANCES OF HAVING MORE CHILDREN WITH ACHROMATOPSIA?

Normally, each gene in the body has two copies or alleles, one allele comes from the mother and the other allele comes from the father. Achromatopsia is an autosomal recessive disorder, which means that for the disease to develop, the two copies or alleles of the gene should be mutated. A carrier is an individual who only has one copy or allele of the mutated gene and does not have achromatopsia. For achromatopsia to develop, a person must inherit one allele of the mutated gene from each parent. This means that both parents must be either carriers or have the disease involving mutations in the same achromatopsia gene. If both parents are carriers, each sibling of an affected individual

with achromatopsia has a 25% chance of having the disease, a 50% chance of being an asymptomatic carrier, and a 25% chance of not having the disease and not being a carrier.



Fig. 1 Child with achromatopsia outdoor



Fig. 2 Same child indoors in dim light

WHAT TREATMENT IS AVAILABLE FOR ACHROMATOPSIA?

Currently there is no cure for achromatopsia. Several clinical trials for gene replacement therapy for CNGA3 and CNGB3-related achromatopsia are currently ongoing and recruiting patients. There are also clinical observational trials that are recruiting patients for clinical evaluation to study the natural history of achromatopsia. Animal models of achromatopsia in dogs and mice have shown promising results in restoring some cone function in the retina. Search [ClinicalTrials.gov](https://clinicaltrials.gov) in the US and [ClinicalTrialsRegister.eu](https://clinicaltrialsregister.eu) in Europe for more information on clinical studies for achromatopsia. Children should be checked for refractive errors (need for glasses). Prescribing glasses to correct refractive conditions such as far-sightedness (hyperopia), near-sightedness (myopia) and astigmatism can improve the vision somewhat but will not restore normal levels of



vision. Red colored or darkly tinted lenses help reducing the sensitivity to light and thus enhance visual functioning. NoIR (injection-molded) plastic wrap-around glasses have a top 'shield' that covers the top of a prescription frame as well as broad side shields which is important since light coming in from any direction around the glasses can be very uncomfortable. Examples are Corning Lenses: CPF 550 lenses (5% transmission, darkened) and CPF 550XD lenses (4% transmission, darkened). These lenses are available through Winchester Optical. There are small, handheld color detecting devices such as Colorino that allow detection of color for specific activities. A newer device known as the Eyeborg system can help people with no color vision to perceive color through sound waves. Artist Neil Harbisson who suffered from achromatopsia was one of the first to use this device. Consultation with low vision services would be valuable and often recommended to find appropriate low vision aids for school, work, or daily activities of living.

HOW WILL THIS CONDITION AFFECT MY CHILD'S SCHOOLING?

With adequate help from teachers for the visually impaired, children with achromatopsia are usually able to attend mainstream schools. Front seat placement, large print books, and magnifying devices can be very helpful. A low vision evaluation will be necessary before school begins. More severely affected individuals may benefit from services available from the State Commission for the Blind and in schools specifically designated for the visually impaired.

IS THERE AN ACHROMATOPSIA SUPPORT GROUP?

- [The Achromatopsia Group](#)

Updated 03/2022