achromatopsia

Achromatopsia is a condition characterized by a partial or total absence of color vision. People with complete achromatopsia cannot perceive any colors; they see only black, white, and shades of gray. Incomplete achromatopsia is a milder form of the condition that allows some color discrimination.

Achromatopsia also involves other problems with vision, including an increased sensitivity to light and glare (photophobia), involuntary back-and-forth eye movements (nystagmus), and significantly reduced sharpness of vision (low visual acuity). Affected individuals can also have farsightedness (hyperopia) or, less commonly, nearsightedness (myopia). These vision problems develop in the first few months of life.

Achromatopsia is different from the more common forms of color vision deficiency (also called color blindness), in which people can perceive color but have difficulty distinguishing between certain colors, such as red and green.

Frequency

Achromatopsia affects an estimated 1 in 30,000 people worldwide. Complete achromatopsia is more common than incomplete achromatopsia.

Complete achromatopsia occurs frequently among Pingelapese islanders, who live on one of the Eastern Caroline Islands of Micronesia. Between 4 and 10 percent of people in this population have a total absence of color vision.

Genetic Changes

Achromatopsia results from changes in one of several genes: *CNGA3*, *CNGB3*, *GNAT2*, *PDE6C*, or *PDE6H*. A particular *CNGB3* gene mutation underlies the condition in Pingelapese islanders.

Achromatopsia is a disorder of the retina, which is the light-sensitive tissue at the back of the eye. The retina contains two types of light receptor cells, called rods and cones. These cells transmit visual signals from the eye to the brain through a process called phototransduction. Rods provide vision in low light (night vision). Cones provide vision in bright light (daylight vision), including color vision.

Mutations in any of the genes listed above prevent cones from reacting appropriately to light, which interferes with phototransduction. In people with complete achromatopsia, cones are nonfunctional, and vision depends entirely on the activity of rods. The loss of cone function leads to a total lack of color vision and causes the other vision problems. People with incomplete achromatopsia retain some cone function. These individuals have limited color vision, and their other vision problems tend to be less severe.

Some people with achromatopsia do not have identified mutations in any of the known genes. In these individuals, the cause of the disorder is unknown. Other genetic factors that have not been identified likely contribute to this condition.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- achromatism
- rod monochromatism
- total color blindness

Diagnosis & Management

These resources address the diagnosis or management of achromatopsia:

- GeneReview: Achromatopsia
  https://www.ncbi.nlm.nih.gov/books/NBK1418
- Genetic Testing Registry: Achromatopsia
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0152200/
- MedlinePlus Encyclopedia: Color Vision Test
  https://medlineplus.gov/ency/article/003387.htm

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Color Blindness
  https://medlineplus.gov/ency/article/001002.htm
- Encyclopedia: Color Vision Test
  https://medlineplus.gov/ency/article/003387.htm
- Health Topic: Color Blindness
  https://medlineplus.gov/colorblindness.html

Genetic and Rare Diseases Information Center

- Achromatopsia 2
  https://rarediseases.info.nih.gov/diseases/9649/achromatopsia-2
- Achromatopsia 3
  https://rarediseases.info.nih.gov/diseases/9650/achromatopsia-3
- Blue cone monochromatism
  https://rarediseases.info.nih.gov/diseases/917/blue-cone-monochromatism

Educational Resources

- American Association for Pediatric Ophthalmology and Strabismus
  https://aapos.org/terms/conditions/10
- American Optometric Association
  http://www.aoa.org/patients-and-public/eye-and-vision-problems/glossary-of-eye-and-vision-conditions/color-deficiency
- Disease InfoSearch: Achromatopsia
  http://www.diseaseinfosearch.org/Achromatopsia/9534
- Florida State University: Human Vision and Color Perception
  http://micro.magnet.fsu.edu/primer/lightandcolor/humanvisionintro.html
- MalaCards: achromatopsia
  http://www.malacards.org/card/achromatopsia
- Orphanet: Achromatopsia
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=49382
- The Pingelapese Colorblind (Genome News Network, June 30, 2000)
  http://www.genomenewsnetwork.org/articles/06_00/pingelapese_colorblind.shtml

Patient Support and Advocacy Resources

- Achromatopsia.info
  http://www.achromatopsia.info/
- Fighting Blindness (Ireland)
  https://www.fightingblindness.ie/
- Low Vision Gateway
  http://www.lowvision.org/achromatopsia_and_color_blindnes.htm
- Prevent Blindness
  http://www.preventblindness.org/color-blindness
- Resource List from the University of Kansas Medical Center: Blind / Visual Impairment
  http://www.kumc.edu/gec/support/visual.html

GeneReviews

- Achromatopsia
  https://www.ncbi.nlm.nih.gov/books/NBK1418

Genetic Testing Registry

- Achromatopsia
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0152200/
- Achromatopsia 2
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1857618/
- Achromatopsia 3
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1849792/
- Achromatopsia 4
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C1841721/
- Achromatopsia 5
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C2751309/
- Achromatopsia 6
  https://www.ncbi.nlm.nih.gov/gtr/conditions/CN158542/

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22achromatopsia%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28achromatopsia%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- ACHROMATOPSIA 2
  http://omim.org/entry/216900
- ACHROMATOPSIA 3
  http://omim.org/entry/262300
- ACHROMATOPSIA 4
  http://omim.org/entry/613856
- CONE DYSTROPHY 4
  http://omim.org/entry/613093
- RETINAL CONE DYSTROPHY 3A
  http://omim.org/entry/610024

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