Blue Cone Monochromacy

What is Blue Cone Monochromacy (BCM)? BCM is a rare genetic condition that is present at birth and causes an inability to perceive red or green colors. It affects about 1 in 100,000 males, and 1 in 10 million females.

Signs and Symptoms Getting a Diagnosis

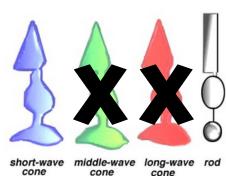
- Shaking or jerking of eyes (nystagmus)
- · Sensitivity to or difficulty seeing in bright light, squinting
- Poor ability to tell colors apart
- Impaired vision
- Nearsightedness

- 1. Eye exam. An eye doctor can perform vision tests to diagnose BCM.
- 2. Genetic testing. If you have symptoms or a family history of BCM, see a genetic counselor to discuss the genetic cause of BCM.

How BCM Changes Vision

Our eyes sense light through cells at the back of the eye called *rods* and *cones*. Rods help with low-light, motion detection, and night vision. Cones help with seeing our sharpest vision in daylight and telling differences between colors.

Red cones, green cones, and blue cones work together so we can see all the colors of the rainbow. In BCM, red and green cones do not work, so people with BCM have trouble telling colors apart. Reds often appear black, and green can appear yellow. They see blue colors best. People with BCM may not see color the same way as someone with normal color vision.



In BCM, only rods (night vision) and blue cones work. People with BCM are often sensitive to bright light and have trouble seeing details.

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Genetics of BCM

- BCM is inherited from the mother and usually only affects males.
- A DNA change, called a genetic variant, causes the red and green cones not to work. The most common mutations happen in genes called OPN1LW, OPN1MW, or a nearby location called the LCR.
 These are all on the X chromosome.
- BCM rarely affects females. If there is a Unaffected daughter Affected daughter of carrier daughter son one X chromosome, the other X chromosome will make up for it. But females can be "carriers" of BCM. They can pass down the genetic variant to their children, even if they have normal vision.
- Males have only one X chromosome. If males inherit a genetic variant, then they will have BCM. Males can pass on their genetic variant to daughters only.

Frequently Asked Questions

Does BCM cause complete blindness? BCM rarely causes complete loss of vision. Vision usually stays the same throughout life. Decreased central vision may occur in some adults as they get older due to macular degeneration. See your eye doctor regularly to check for changes in the macula.

How does BCM impact daily living? People with BCM can live full, productive lives. They will need special accommodations for low vision, such as school accommodations, low vision assistive devices, and sports adaptations. Some may be legally blind, but others may be able to get a driver's license with a low vision assistive device. They may need to be more careful of their physical surroundings and wear sunglasses to reduce brightness.

Is there treatment for BCM? There is no treatment for BCM. Talk to your eye doctor to discuss changes in vision and use of special glasses, contacts, or low vision assistive devices.

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More Support and Information

<u>Cincinnati Association for the Blind & Visually Impaired</u>
https://www.cincyblind.org/

<u>Cincinnati Children's Low Vision Program</u>

https://www.cincinnatichildrens.org/service/o/ophthalmology/services/

vision-rehabilitation

Clovernook Low Vision Clinic

https://clovernook.org/youth-services/low-vision-clinic/

BCM Families Foundation

https://www.blueconemonochromacy.org/

The Low Vision Centers of Indiana

http://www.eyeassociates.com/

Foundation Fighting Blindness

https://www.fightingblindness.org/

Achromatopsia

http://www.achromatopsia.info/blue-cone-monochromatism/