

OPN1MW Gene

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1. Introduction

The *OPN1MW* gene provides instructions for making a protein that is essential for normal color vision. This protein is found in 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, that transmit visual signals from the eye to the brain. Rods provide vision in low light. Cones provide vision in bright light, including color vision. There are three types of cones, each containing a specific pigment (a photopigment called an opsin) that is most sensitive to particular wavelengths of light.

The *OPN1MW* gene provides instructions for making an opsin pigment that is more sensitive to light in the middle of the visible spectrum (yellow/green light). Cones with this pigment are called middle-wavelength-sensitive or M cones. In response to light, the photopigment triggers a series of chemical reactions within an M cone. These reactions ultimately alter the cell's electrical charge, generating a signal that is transmitted to the brain. The brain combines input from all three types of cones to produce normal color vision.

People can have one or more copies of the *OPN1MW* gene in each cell. All copies of this gene are located in a row on the X chromosome near another opsin pigment gene, *OPN1LW*. The *OPN1LW* gene provides instructions for making a photopigment that is more sensitive to light at long wavelengths (in the orange/red part of the visible spectrum). A nearby region of DNA, known as the locus control region (LCR), regulates the activity of the *OPN1MW* and *OPN1LW* genes. Only the two opsin pigment genes nearest the LCR, generally the *OPN1LW* gene and the first copy of the *OPN1MW* gene, are active in the retina and contribute to color vision.

2. Health Conditions Related to Genetic Changes

2.1. Color vision deficiency

Several kinds of genetic changes involving the *OPN1MW* gene cause red-green color vision defects, a form of color vision deficiency that makes it difficult or impossible to distinguish between shades of red, yellow, and green. Most red-green color vision defects result from structural rearrangements involving the *OPN1LW* and *OPN1MW* genes. Because these genes are so similar, they occasionally swap genetic material when the genes are being passed from parent to child. This swapping, called recombination, can ultimately delete genetic material from one or both genes or lead to the formation of a hybrid pigment gene that contains part of the *OPN1MW* gene and part of the *OPN1LW* gene.

Less commonly, red-green color vision defects can result from an *OPN1MW* gene mutation that changes a single protein building block (an amino acid) in the middle-wave-sensitive photopigment. This mutation replaces the amino acid cysteine with the amino acid arginine at position 203 (written as Cys203Arg or C203R).

When *OPN1MW* gene mutations lead to completely nonfunctional M cones, color vision depends entirely on the other two types of cones. The specific type of red-green color vision deficiency that results from a total loss of M cone function is called deuteranopia. A less severe red-green color vision defect called deuteranomaly occurs when a partially functional hybrid pigment gene replaces the normal *OPN1MW* gene. The photopigments made from these hybrid genes usually have abnormal visual properties that impair red-green color vision.

A rarer form of color vision deficiency, blue cone monochromacy, severely reduces sharpness of vision (visual acuity) and affects the ability to perceive most colors. This condition also includes other vision problems that are not typically found with red-green color vision defects. Blue cone monochromacy occurs when genetic changes prevent the opsin pigments

produced from both the *OPN1MW* and *OPN1LW* genes from functioning normally. In some cases, the condition is caused by a deletion of the LCR, which normally controls the activity of the *OPN1MW* and *OPN1LW* genes. A loss of the LCR prevents the production of pigments from both genes. As a result, people with this condition have only functional cones with short-wavelength-sensitive photopigment (S cones), which leads to reduced visual acuity and poor color vision. The cone abnormalities also underlie the other vision problems in people with blue cone monochromacy.

3. Other Names for This Gene

- CBBM
- GCP
- green cone photoreceptor pigment
- green cone pigment
- green-sensitive opsin
- M-pigment
- middle-wave-sensitive pigment
- OPN1MW1
- OPSG_HUMAN
- opsin 1 (cone pigments), medium-wave-sensitive
- photopigment apoprotein

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