OPN1LW gene

opsin 1, long wave sensitive

Normal Function

The *OPN1LW* 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 *OPN1LW* gene provides instructions for making an opsin pigment that is more sensitive to light in the yellow/orange part of the visible spectrum (long-wavelength light). Cones with this pigment are called long-wavelength-sensitive or L cones. In response to light, the photopigment triggers a series of chemical reactions within an L 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.

The *OPN1LW* gene is located next to another opsin pigment gene, *OPN1MW*, on the X chromosome. The *OPN1MW* gene provides instructions for making a photopigment that is more sensitive to light at middle wavelengths (yellow/green light). Most people have one copy of the *OPN1LW* gene and one or more copies of the *OPN1MW* gene on each X chromosome. A nearby region of DNA, known as the locus control region (LCR), regulates the activity of these 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.

Health Conditions Related to Genetic Changes

<u>color vision deficiency</u>

Several kinds of genetic changes involving the *OPN1LW* 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 *OPN1LW* gene

and part of the *OPN1MW* gene. Less commonly, red-green color defects can result from changes in single DNA building blocks (base pairs) in the *OPN1LW* gene.

When *OPN1LW* gene mutations lead to completely nonfunctional L 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 L cone function is called protanopia. A less severe red-green color vision defect called protanomaly occurs when a partially functional hybrid pigment gene replaces the normal *OPN1LW* gene. The photopigments made from these hybrid genes usually have abnormal visual properties that impair red-green color vision.

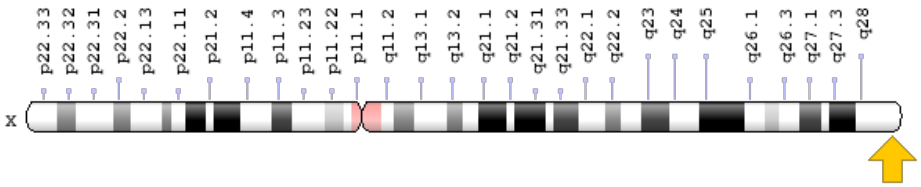
A common variation (polymorphism) in the *OPN1LW* gene accounts for subtle differences in normal color vision. This change alters a single protein building block (amino acid) in the resulting photopigment, replacing the amino acid serine with the amino acid alanine at position 180 (written as Ser180Ala). Researchers suggest that the Ser180Ala polymorphism also plays a role in determining the severity of color vision loss in people with red-green color vision defects.

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.

Chromosomal Location

Cytogenetic Location: Xq28, which is the long (q) arm of the X chromosome at position 28

Molecular Location: base pairs 154,144,224 to 154,159,032 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- L-pigment
- long-wave-sensitive pigment
- opsin 1 (cone pigments), long-wave-sensitive
- OPSR_HUMAN
- RCP
- red cone photoreceptor pigment
- red cone pigment
- red-sensitive opsin

Additional Information & Resources

<u>Educational Resources</u>

- Florida State University: Human Vision and Color Perception
  http://micro.magnet.fsu.edu/primer/lightandcolor/humanvisionintro.html
- Neuroscience (second edition, 2001): Cones and Color Vision
  https://www.ncbi.nlm.nih.gov/books/NBK11059/
- Webvision: The Organization of the Retina and Visual System (updated 2009): Color Vision
  https://www.ncbi.nlm.nih.gov/books/NBK11537/

GeneReviews

- Red-Green Color Vision Defects
  https://www.ncbi.nlm.nih.gov/books/NBK1301

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28OPN1LW%5BTIAB%5D%29+OR+%28%28red+cone+pigment%5BTIAB%5D%29+OR+%28Red-sensitive+opsin%5BTIAB%5D%29+OR+%28L+pigment%5BTIAB%5D%29%29+OR+%28%28RCP%5BTIAB%5D%29+AND+%28vision%5BTIAB%5D%29%29+OR+%28%28opsin%5BMAJR%5D%29+AND+%28red%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- OPN1LW AND OPN1MW GENES, CONTROLLER OF
  http://omim.org/entry/300824
- OPSIN 1, LONG-WAVE-SENSITIVE
  http://omim.org/entry/300822

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_OPN1LW.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=OPN1LW%5Bgene%5D
- HGNC Gene Family: Opsin receptors
  http://www.genenames.org/cgi-bin/genefamilies/set/215
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=9936
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5956

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