OPN1SW gene

opsin 1, short wave sensitive

Normal Function

The *OPN1SW* 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 *OPN1SW* gene provides instructions for making an opsin pigment that is more sensitive to light in the blue/violet part of the visible spectrum (short-wavelength light). Cones with this pigment are called short-wavelength-sensitive or S cones. In response to light, the photopigment triggers a series of chemical reactions within an S 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.

Health Conditions Related to Genetic Changes

Color vision deficiency

At least six mutations in the *OPN1SW* gene have been found to cause a relatively uncommon form of color vision deficiency called blue-yellow color vision defects or tritan defects. These defects cause problems with differentiating shades of blue and green and cause difficulty distinguishing dark blue from black. Each of the known *OPN1SW* gene mutations changes a single protein building block (amino acid) in the short-wave-sensitive photopigment, which causes the photopigment to be partially or totally nonfunctional. Researchers suggest that S cone cells without functional photopigment die prematurely or cannot transmit visual signals to the brain normally.

When *OPN1SW* gene mutations lead to completely nonfunctional S cones, color vision depends entirely on the other two types of cones. The specific type of blue-yellow color vision deficiency that results from a total loss of S cone function is called tritanopia. A less severe blue-yellow color vision defect called tritanomaly occurs when S cones function abnormally.

Other Names for This Gene

- BCP
- blue cone photoreceptor pigment
- blue cone pigment
- blue-sensitive opsin
- BOP
- OPSB_HUMAN
- opsin 1 (cone pigments), short-wave-sensitive
- S-pigment
- short-wave-sensitive pigment

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of OPN1SW (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=611[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28OPN1SW%5BTIAB%5D%29+OR+%28%28blue+cone+pigment%5BTIAB%5D%29+OR+%28short-wave-sensitive+pigment%5BTIAB%5D%29+OR+%28Blue-sensitive+opsin%5BTIAB%5D%29%29+OR+%28%28S+cone%5BTIAB%5D%29+AND+%28tritan%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

Catalog of Genes and Diseases from OMIM

- OPSIN 1, SHORT-WAVE-SENSITIVE; OPN1SW (https://omim.org/entry/613522)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/611)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=OPN1SW[gene])

References

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Genomic Location

The *OPN1SW* gene is found on chromosome 7 (https://medlineplus.gov/genetics/chromosome/7/).

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