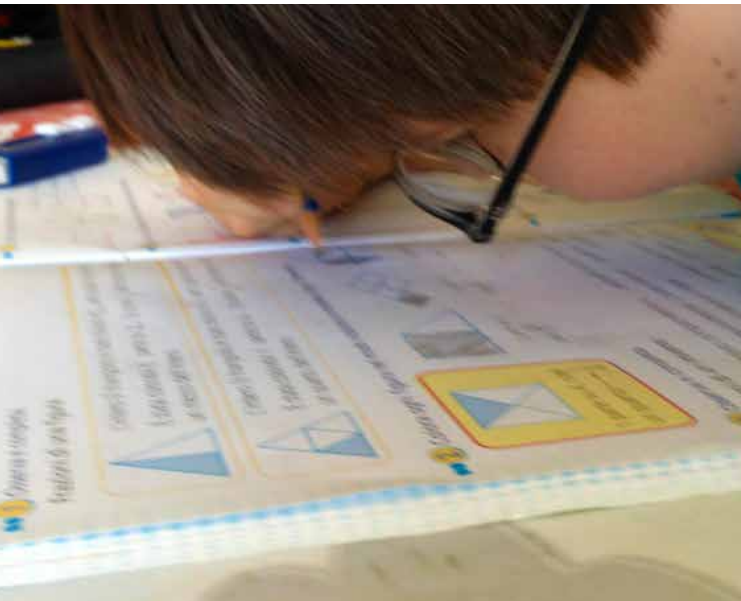


The BCM Families Foundation

BCM Families Foundation is a non-profit organization based in the United States that was established in 2014 by families touched by the rare and neglected disease named Blue Cone Monochromacy (BCM).

The Mission

The mission of the BCM Families Foundation is to eradicate Blue Cone Monochromacy by supporting the most promising **biomedical** and **scientific research** that will ultimately lead to **a cure**



BCM Families Foundation is a tax-exempt charity under IRS Code, Section 501(c)(3)

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#Cure4BCM

The BCM C198R mouse

a novel experimental model for
Blue Cone Monochromacy



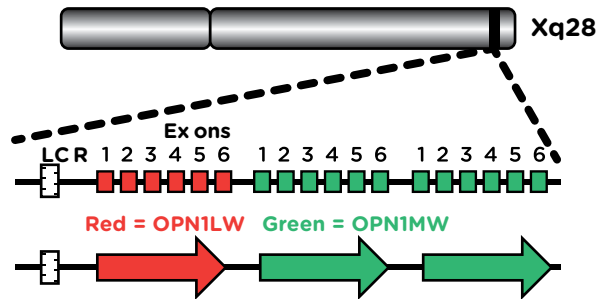
Blue Cone Monochromacy

Blue Cone Monochromacy (BCM) is a rare X-linked disease of the retina due to cone photoreceptors dysfunction that causes color vision abnormalities, strong glare, nystagmus and reduced visual acuity. It affects approximately 1 in 100,000 individuals. There are three types of cone photoreceptors in the human retina that express different photopigments (opsins) enabling color vision.

Cell expressed	Opsin Gene	Chromosome
L-cone (Red)	OPN1LW	Xq28
M-cone (Green)	OPN1MW	Xq28
S-cone (blue)	OPN1SW	7q31.3-32

Blue Cone Monochromacy is caused by simultaneous mutations in the OPN1LW (LW) and OPN1MW (MW) genes, that encode for the long and middle wave length sensitive cone opsins respectively.

OPN1LW and OPN1MW are located in a head-to-tail arrangement on chromosome Xq28.



Deleterious mutations include deletions of the locus control region (LCR), genomic rearrangements and point mutations. **The most common point mutation is a substitution of cytosine for thymine at nucleotide position 1101, which corresponds to a substitution of arginine for cysteine at amino acid position 203 (C203R) and may result in a misfolded, non functional opsin protein.**

BCM C198R Mouse

So far, BCM has been investigated in models such as knockout mice that mimic large gene deletions seen in humans. Until recently, there was not a preclinical model for the common BCM C203R point mutation which is thought to cause a possibly different pathomechanism due to protein misfolding.

With an aim of accelerating the study, identification and development of treatments that could be effective for the people carrying this specific mutation, the BCM Families Foundation embarked on the creation of the respective mouse model: **the BCM C198R mouse mutant** was created at Charles River Laboratories and then donated to The Jackson Laboratory.

How to request the BCM C198R Mouse

The BCM C198R mouse model is available at The Jackson Laboratory as Stock No. 031385.

If you are interested in the strain, please visit the website at www.jax.org and register your interest or contact the Customer Service to be informed about availability

Genetic engineering

The mutation was introduced into the mouse MW opsin gene via in vivo gene editing using CRISPR/Cas9 technology causing a p.C198R in the mouse MW opsin which corresponds to p.C203R in the human LW/MW opsin.

