BCM FAMILIES FOUNDATION

28 February 2023

Rare Disease Day

RAREDISEASEDAY.ORG

Getting to know Blue Cone Monochromacy

Dear ophthalmologists, low-vision specialists, optometrists, geneticists, pediatricians, other medical professionals, and friends of BCM:

28 February 2023 marks the sixteenth international Rare Disease Day coordinated by European Organisation for Rare Diseases (EURORDIS) and National Organization for Rare Disorders (NORD).

Today patients, families, and volunteers of BCM Families Foundation reached you with this flyer because we would like to tell you about Blue Cone Monochromacy, also known as BCM, a rare genetic eye condition affecting 1 in 100,000 people.

BCM Families Foundation is a non-profit 501(c)(3) organization, founded and registered in the US by patients affected by BCM and their relatives. Our mission is to cure Blue Cone Monochromacy.

BCM at a glance

Blue Cone Monochromacy is a rare retinal disorder and is inherited in an X-linked manner.
Affected males commonly show symptoms soon after birth and BCM is thought to be a relatively stable disease, affecting an individual for his whole life.
Female carriers rarely show symptoms, and when they do, they are very mild.

Symptoms of BCM

Poor visual acuity ranging from 20/60 to 20/200 or greater, in some cases legal blindness.

Poor or no color discrimination, Patients with BCM have functioning rod photoreceptors and blue cone photoreceptors. However, they lack function of their red (L) and green (M) cone photoreceptors.

Hemeralopia, Patients with BCM have severe difficulty coping with glare because they only have blue cone function at higher light levels. The severity of the symptoms, commonly known as photophobia, varies by the integrity of remaining blue cone cells.

Nystagmus: Nystagmus is usually present from about 3-6 months of age. It is usually, but not always, pendular and may decrease with age.

Myopia or **high myopia**

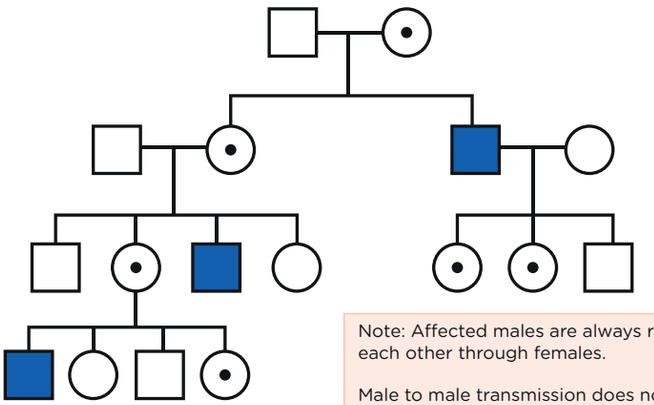
X-linked inheritance family pedigree. Mutations causing BCM are located in the X chromosome in the following genes and supporting components: **OPN1LW, OPN1MW** and the upstream **LCR** (Locus Control Region). Deletion in the LCR or a point mutation C203R on a single L-M opsin gene are most common forms.

So if you see a 6 month old boy with nystagmus and the parents speak about another male in their family (on the mother's side) with similar eye problems, please consider a diagnosis of BCM.

X-Linked recessive Pedigree

Key

- = Male affected
- = Male unaffected
- = Female carrier
- = Female non-carrier



Note: Affected males are always related to each other through females.

Male to male transmission does not occur.

For confirmatory diagnosis, please conduct genetic testing at:

University of Tuebingen
Molecular Genetics Laboratory - Germany
Contact: Susanne Kohl, Msc, PhD
Email: susanne.kohl@uni-tuebingen.de
BCMFF Collaboration

Radboud Medical Genetics Institute
Nijmegen The Netherland
Email: info@gdnm.nl
Web: https://order.radboudumc.nl

Molecular Vision Laboratory
1920 NE Stucki Ave, Suite 150 Hillsboro, OR 97006
Contact: John Chiang, PhD, FACMG
Email: jchiang@mvisionlab.com
Web: www.molecularvisionlab.com

John and Marcia Carver Nonprofit Genetic Testing Laboratory University of Iowa
375 Newton Rd. 4111 MERF Iowa City, IA 52242
Email: carverlab@uiowa.edu
Web: https://www.carverlab.org

Participate in the research supported by BCM Families Foundation at:

University of Pennsylvania - Dr. S.G. Jacobson **BCMFF Collaboration**
https://www.pennmedicine.org/providers/profile/samuel-jacobson

Medical College of Wisconsin - Dr. J. Carroll
Web: https://www.mcw.edu/departments/ophthalmology-eye-institute/research/advanced-ocular-imaging-program

For more information about the disease, we invite you to visit the Online Mendelian Inheritance in Man (OMIM) page for BCM (http://omim.org/entry/303700) and our website (www.bcmfamilies.org)

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