BCM FAMILIES FOUNDATION

28 February 2019

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 2019 marks the twelfth international Rare Disease Day coordinated by European Organisation for Rare Diseases (EURORDIS) and National Organization for Rare Disorder (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 eradicate Blue Cone Monochromacy.

BCM at a glance

Blue Cone Monochromacy is a rare genetic X-linked retinal disorder. Affected males commonly show symptoms soon after birth, while females carriers only rarely show some of the symptoms in a mild form. BCM is a stable disease, affecting an individual for his whole life.

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, BCM individuals have only rods photoreceptors and blue cones photoreceptors. They lack the photopigments in the red (L) and the green (M) cones.

Hemeralopia, BCM individuals have severe difficulty coping with glare because only blue cones and rods are available to function in bright light. The severity of the symptoms, commonly know as photophobia, varies by the integrity of remaining blue cone cells.

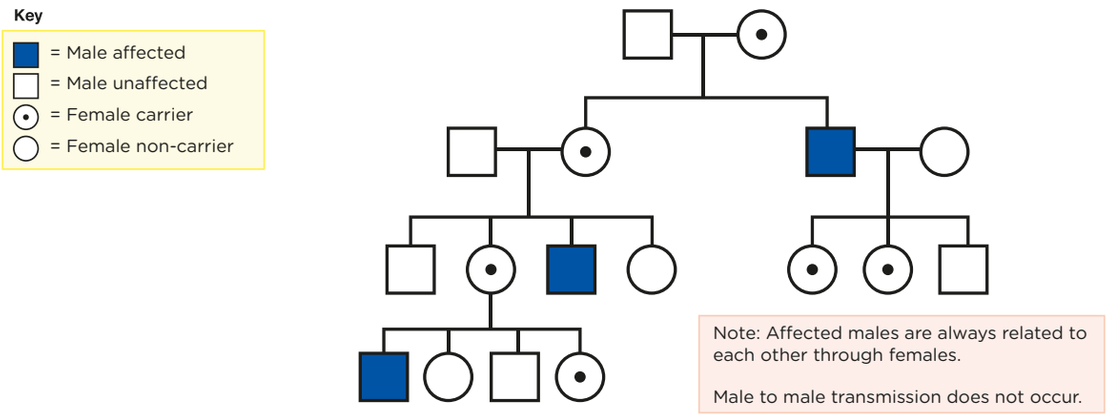
Nystagmus: Nystagmus is usually present from about 3-6 months of age. It is usually, but not always, pendular and may decrease with age.

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



For confirmatory diagnosis, please conduct genetic testing and participate in the research supported by BCM Families Foundation at:

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

Another laboratory that offers the BCM DNA test is:

Molecular Vision Laboratory
Biomedical Research Building FL2
3181 SW Sam Jackson Park Rd. Portland, OR 97239-3098 USA
Contact: Director John (Pei-Wen) Chiang, PhD, FACMG – Director
Email: JChiang@mvisionlab.com

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)

Thank you

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