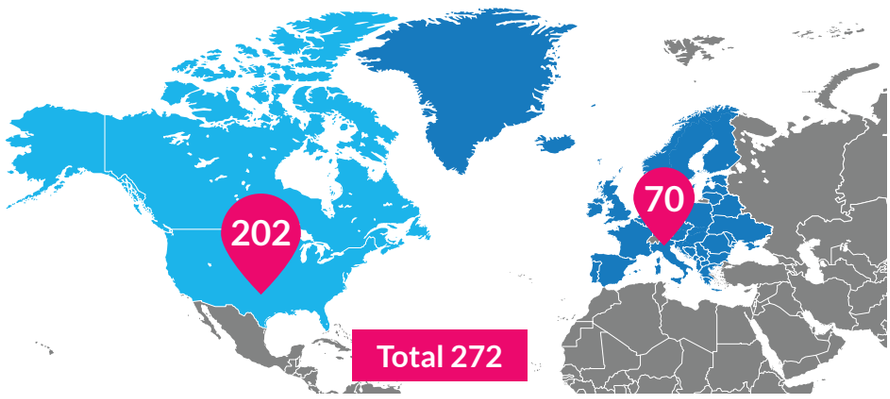
The BCM Patient Registry

An International Patient Registry for Blue Cone Monochromacy

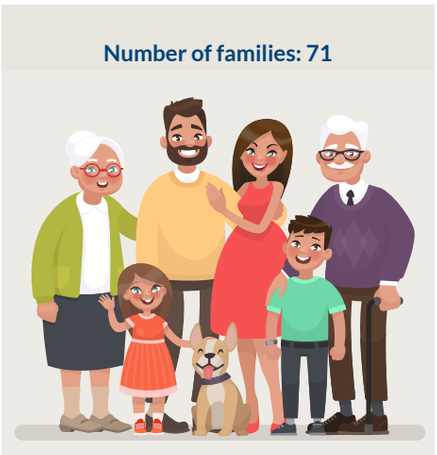
www.BCMRegistry.org

Map of countries that contributed to 2019-2023 data



North America (USA, Canada) 74% | Europe+UK 26%

Families



Adults & Children*

Under 6	6%
6-12	16%
12-18	24%
18-30	21%
30-40	5%
40-50	13%
Over 50	15%

Adults 54% | Minors 46%

Average number of living people with BCM in a family **3.8**

Average number of total people with BCM in a family **5.1**

What is the Blue Cone Monochromacy International Patient Registry?

The BCM Registry is a list of men & boys diagnosed with Blue Cone Monochromacy, an inherited retinal disease. They consent to be registered and to share their clinical and genetic data. BCM Registry is made to increase understanding of BCM by providing doctors and researchers with first-hand information about the people with this disease, both individually and as a group. The BCM Registry can capture the demographics of this disease, identify patients for clinical trials, track clinical outcomes, and more.

Purpose

The Purpose of the Registry is to collect data over time from patients with BCM from all over the world. The Registry aims to increase knowledge about:

- Clinical features of BCM;
- Natural course of BCM;
- Long-term outcomes of BCM;
- And ultimately help find a cure for BCM.

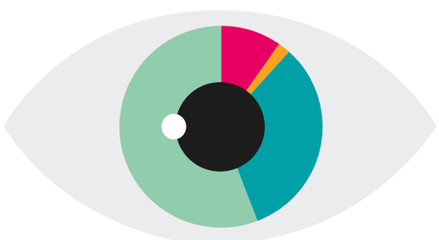
Mission

This BCM Registry® is an online Patient Registry dedicated to a rare genetic retinal disease, Blue Cone Monochromacy. The Registry was created by BCM Families Foundation, a 501(c)(3) non-profit patient-led organization with the mission to cure BCM.

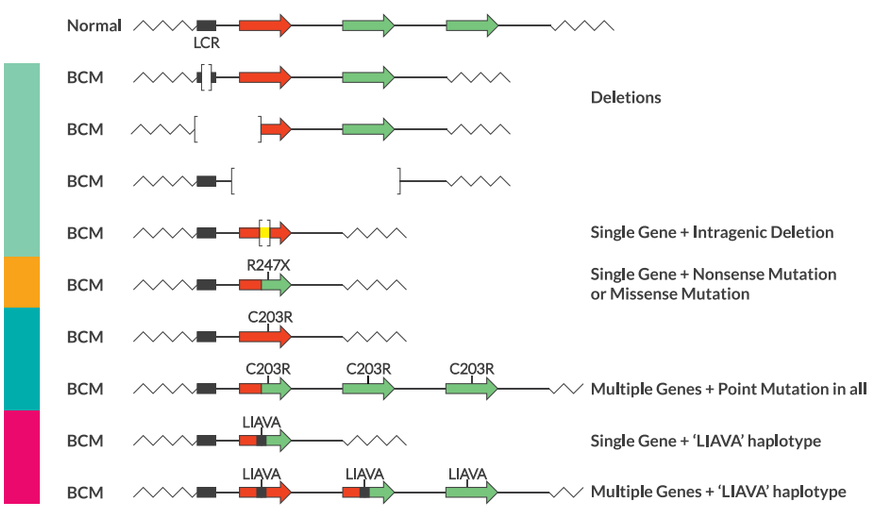
* Data about age are only about BCM patients who enrolled to the Registry and not about their affected relatives

Causative mutations (Data about mutations are about BCM patients who enrolled and inserted their DNA test report plus all their alive affected relatives).

This picture was kindly provided by Prof. Bernd Wissinger, University of Tübingen, Germany



Deletions - 55,1% | Other PT - 1,1%
C203R - 37,9% | Exon 3 - 5,9%



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www.BCMRegistry.org

Participating Clinicians

USA

- **Tomas S. Aleman**, M.D., Director Center for Hereditary Retinal Degenerations and Retinal Function Department, Scheie Eye Institute, University of Pennsylvania
- **Alessandro Iannaccone**, M.D., M.S., Astellas Pharma US, and University of North Carolina, School of Medicine, Chapel Hill, NC
- **Joseph Carroll**, Ph.D., Professor of Ophthalmology & Visual Sciences, Biophysics, and Cell Biology, Neurobiology and Anatomy, Medical College of Wisconsin, Milwaukee, WI
- **Amy K. Hutchinson**, M.D., Professor of Ophthalmology, Emory Eye Center, Emory University, Atlanta, GA
- **David Birch**, Ph.D., Retina Foundation of the Southwest, Dallas, TX.
- **Jacque Duncan**, M.D., Professor, Ophthalmology, School of Medicine, UCSF, San Francisco, CA
- **Marc Mathias**, M.D., University of Colorado, School of Medicine, Aurora, CO

Europe

- **Michel Michaelides**, M.D., Professor of Ophthalmology, Moorfields Eye Hospital, London, United Kingdom
- **Lucia Ziccardi**, M.D., Ph.D., GB Bietti Foundation, Rome, Italy
- **Prof. Dr. med. Katarìna Štingl**, Ophthalmologist, University of Tübingen, Centre for Ophthalmology, Tübingen, Germany
- **Elisabetta Martina**, M.D., IRCCS - San Raffaele Scientific Institute, Milan, Italy
- **Cristiana A. Marchese,** Ph.D., Retina Italia, Torino, Italy
- **Dr. Pierre Bitoun**, Groupe Médical Jarente , Paris
- **Dr. Cristina Irigoyen Laborra**, CSUR Inherited Retinal Disease Coordinator, Spain

What information is collected in the BCM Patient Registry ?

The Registry collects the following data:

- Personal information from individuals with BCM including, but not limited to, name, date of birth, city of birth, country of birth, address, phone number, and email address;
- Information about diagnosis of BCM including genetic test, vision tests, and exam results;
- Information about personal and family health history as it relates to BCM;
- The data is not identifiable and stored in a secure encrypted database.

Data analysis method

In this poster, we have aggregated the data of those who participated in the BCM Patient Registry. Since the participants are self-selected volunteers, they do not represent the general population of BCM males and their relatives. The aggregate disproportionally excludes BCM families without access to diagnosis or to computers or high-speed internet services.

Only sums of data are offered. The Patient Registry is not a random sample. Thus, tests of statistical significance do not apply.

BCM FAMILIES FOUNDATION
www.blueconemonochromacy.org

BCM Families Foundation is a non-profit organization incorporated in May 2014 in USA, in the state of Delaware, by families affected by Blue Cone Monochromacy (BCM). BCM Families Foundation is a tax-exempt charity under IRS Code, Section 501 (c)(3).

The mission of BCM Families Foundation is to cure Blue Cone Monochromacy by supporting the most promising biomedical and scientific research.

Project made with Otto per mille della Chiesa Valdese funds

Otto per mille CHIESA VALDESE
UNIONE DELLE CHIESE METODISTE E VALDESI

How can I participate in the BCM Registry?

Adults and children with BCM may participate in the BCM Registry. You must have genetic confirmation of BCM. During registration, you have to upload your genetic test report.

Enrollment

The BCM Registry is patient-powered. You enroll yourself or your child and select the name of your doctor, who will check and validate all medical data.

Self-Registration

Go to www.BCMRegistry.org and enroll yourself

Full instructions are available at:
www.blueconemonochromacy.org/patient-registry

1
Prepare all documents

2
Create your account at
www.BCMRegistry.org

3
Send an email to:
Registry.Manager@bcmfamilies.org

Download the BCM Registry flyer and give it to your relatives / patients affected by BCM