

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 Director, 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. Katarina 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 disproportionately 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 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

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

The BCM Registry is an online patient registry dedicated to Blue Cone Monochromacy (BCM), established by the BCM Families Foundation (BCMFF). The BCMFF is the only patient-led organization worldwide with a mission to eradicate BCM. Established in 2014 in the United States, the Foundation has gathered a community of more than 300 people (130 families) from around the globe.

BCM affects 1 in 100,000 people, primarily males, who experience loss of visual acuity, photophobia, myopia, nystagmus, impaired color vision from birth and therefore, a significant challenge to quality of life. BCMFF provides research funds and infrastructures that aid discovery of new treatments and improvement of current ones.

By collecting and analyzing many patients' personal and clinical data, the path toward clinical trials of innovative therapies.

The BCM Registry is available at www.BCMRegistry.org, and it BCMFF through the Board of Directors. The Registry is governed by a board of clinicians and researchers, as patient representatives.

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

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

When you join the Registry, with the help of your doctor, you will be connected with those of other people with BCM, creating the diagnosis must be confirmed by genetic testing, and your medical data will be updated by your doctor.