No-Cost DNA Test Program for Blue Cone Monochromacy

BCM FAMILIES FOUNDATION

Universitätsklinikum Tübingen

Why get tested?

If you have a clinical diagnosis of Blue Cone Monochromacy, there are many reasons to test your DNA:

- to have a genetic confirmation of the clinical diagnosis, as clinical diagnosis is often very difficult to reach;
- to understand how the disease spreads within your own family and what are the chances of passing the disease to your children;
- to help scientific research to find all the possible genetic mutations that lead to the disease; this is of fundamental importance to improve the diagnostic tests themselves – to make them accurate and complete;
- for diseases such as Blue Cone Monochromacy, for which gene therapies are being developed, it is important for the patient to know his causative mutation, in order to know if he will be able to have access to that therapy.

The lack of an accurate diagnosis can have far-reaching consequences for patients and their families and for the entire BCM community. If you do not reach the true accurate diagnosis of a rare pathology such as Blue Cone Monochromacy, you cannot support the path toward the cure and identify your global community to achieve together the cure of the genetic disease, given that gene therapies can be different for each gene and also for each causative mutation.

Program Overview

The No-Cost Genetic Testing Program for Blue Cone Monochromacy provides no-cost genetic testing for individuals with a clinical diagnosis of Blue Cone Monochromacy. Targeted familial variant testing is also available to female relatives of individuals who receive a positive result through the program and meet certain criteria.

This testing Program is supported by the **BCM Families Foundation**, a nonprofit organization dedicated to find a cure for Blue Cone Monochromacy. The genetic testing is performed at **Dr. Bernd Wissinger's Lab, University of Tübingen, Germany.**

The efficiency of the test in this Program is very high, greater than 90%, because the Wissinger's Lab continuously updates the list of BCM causative mutations, is able to find all known BCM causative mutations and to perform differential diagnosis with other diseases showing similar clinical features, for example **Achromatopsia**, **Bornholm Eye Disease**, **Cone Dystrophy**. This efficiency is unique and is not found in other laboratories which on the contrary might not test for all BCM causative mutations and therefore could report a null response.

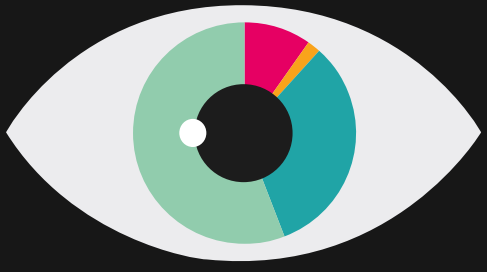
The aim of this program for the BCM Families Foundation is to enlarge inclusion and promote equity to the access to the genetic diagnosis of Blue Cone Monochromacy. The high cost of the test at the very few laboratories that offer it and the lack of information create barriers especially for the most socio-economically disadvantaged individuals, precluding inclusion and diversity in diagnosis.

The only cost to patients is the cost of extracting blood samples and sending them to the laboratory.

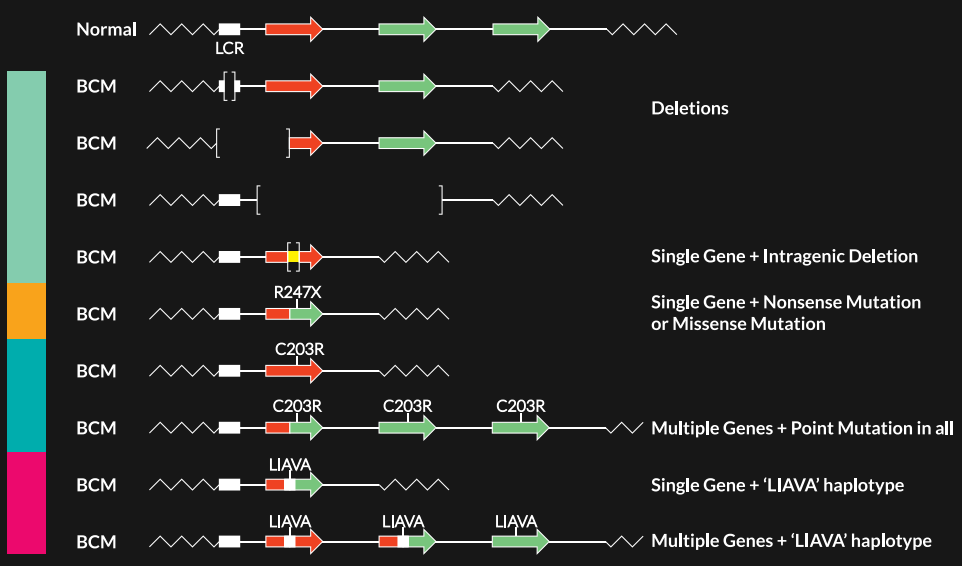
Genes and Causative mutations

X-Chromosome: genes OPN1LW, OPN1MW and the upstream Locus Control Region (LCR).
BCM is a rare genetic disease of the retina caused by genetic mutations on **genes OPN1LW, OPN1MW and the upstream Locus Control Region (LCR).**

This picture was kindly provided by Prof. Bernd Wissinger, University of Tübingen, Germany and percentages come from 2019-2023 BCM Patient Registry aggregated data



Deletions - 55.1%
C203R - 37.9%
Other PT - 1.1%
Exon 3 - 5.9%



Criteria For Test

Participants who undergo genetic testing with the Program must meet these criteria:

- No geographical restrictions
- Have a clinically confirmed diagnosis of Blue Cone Monochromacy and a **Family Pedigree**
- All patients with the genetic confirmation of Blue Cone Monochromacy are encouraged to enroll in the BCM Patient Registry at **www.BCMRegistry.org**

Female relatives who want to test their carrier status must meet these criteria:

- No geographical restrictions
- Have a blood relative tested through the Program who received an informative positive genetic testing result for Blue Cone Monochromacy
- Being age 18+
- Have a family Pedigree
- Targeted testing will be available to X-linked females in the family but not to obligated carriers – that is daughters of an affected male.

Criteria For Clinicians ordering the test

Be willing to join the BCM Registry at **www.BCMRegistry.org** and validate diagnosis of their patients inside the Registry.

Instruction on how to send the blood sample for the DNA test.

https://www.blueconemonochromacy.org/no-cost-dna-test/

Summary

- The No-Cost genetic testing Program for Blue Cone Monochromacy is an open access, no-cost program for patients with a clinical diagnosis of Blue Cone Monochromacy.
- Patients with a clinical diagnosis of Blue Cone Monochromacy can be included in the program.
- Females age 18+ with an affected relative previously tested can test their carrier status, unless this female is the daughter of a father with BCM, therefore an obligate carrier.
- The Program can help eliminate barriers experienced currently by the most socio-economically disadvantaged individuals who are not being diagnosed.
- The Program is supported by the BCM Families Foundation and by selected sponsors.
- The Wissinger Lab at the University of Tübingen in Germany is performing the tests and is able to find all possible Blue Cone Monochromacy causative mutations.
- All patients with the genetic confirmation of Blue Cone Monochromacy are encouraged to enroll in the BCM Patient Registry at **www.BCMRegistry.org**

Contacts

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BCM Families Foundation

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BCM Families Foundation is a 501 (c)(3) charity with the mission to find a cure for Blue Cone Monochromacy

Photo credit to Universitätsklinikum Tübingen (UKT)/Beate Armbruster