



BCM
FAMILIES
FOUNDATION

NEWSLETTER



Newsletter #3 - Holiday Edition

Dear Families and followers,

Another year has passed by, and we would like to share with you the latest news and events that have taken us towards the end of 2016.

- The first animal model for the C203R causative mutation will be developed by BCMFF through a European lab, Charles River



In the past few years BCMFF supported the first scientific research in which a mouse affected by BCM has been treated with gene therapy. For the first time a scientist, Dr. W. W. Hauswirth at University of Florida, developed a BCM-affected mouse without the M opsin. Due to the fact that mice do not have L-opsin, Hauswirth's animal model is very similar to the human BCM case in which the causative mutation is a deletion, that is absence of the L and M opsin proteins inside the retina cones cells.

BCMFF has been helping patients with genetic tests for many years. Statistical data coming from the laboratories report that the C203R mutation is a BCM causative mutation with high frequency. Nobody until now has developed an animal model for this BCM causative mutation although a similar model will be really useful, enabling scientists to test the same gene therapy used for deletions in the C203R case.

BCMFF with the help of the Scientific Advisory Board decided to create an animal model of the C203R mutation. We received quotes from companies and selected a company named Charles River. The cost of this strategic project will be \$ 27,000 and we are looking for a sponsor and donors to help BCMFF with this project.





- Fundraising events - we are looking for volunteers!

BCMFF is going to start a collaboration with an agency in New York for fundraising events and meetings with people able to financially support our mission. People interested in helping with public relations and in representing BCMFF and his mission during meetings are welcome to contact our board. Please send an email to info@bcmfamilies.org with subject of the email: "New York PR"

We would like BCMFF to have a Board of Ambassadors. An Ambassador is a person who can represent plus has the ability to influence, who can use her/his contacts to help and support BCMFF, who is able to gather attention and financial support for our mission.

If you know a person who can become a BCMFF Ambassador please refer her/him to our board. Please send an email to info@bcmfamilies.org



- BCMFF and University of Tübingen finalized an agreement for the study of all the possible BCM causative mutations, particularly LIAVA/MIAVA

University of Tübingen is a very important center in Europe, it's purpose is for the diagnosis and treatment of inherited eye diseases. Not only does this institution keep a biobank of more than

23,000 DNA samples of people affected by inherited eye disease, but also at this university Dr. Susanne Kohl discovered all the causative genes of achromatopsia.

Moreover, in recent years the University of Tübingen started a clinical test of the AAV-based gene therapy for achromatopsia CNGA3, treating the first 9 study participants affected by achromatopsia.

Last November 2016 BCMFF finalized an agreement with University of Tübingen that supported the molecular studies of all the BCM causative mutations, especially the so-called LIAVA/MIAVA mutations, more rare than deletions and point mutations (C203R). The grant provides \$38,000 for 1 year and last November BCMFF delivered \$19,000.

Any help with the support of this project is really welcome. The next instalment of funds will be delivered in April 2017.

Main objectives of the project are:

- Development of sensitive and reliable methods for the detection of mutations and structural aberrations in the opsin gene cluster;
- Development and maintenance of Standardized Operation Procedures for the genetic testing for BCM;
- Reference center for the genetic testing of clinically diagnosed BCM patients;
- Provision of reference material for quality control in genetic testing for BCM;
- Determination of mutation origin and new mutation rates in the opsin gene cluster.

Our aim is to involve University of Tübingen with the creation of a BCM biobank that will be part of our Patient Registry project. Moreover we would like to involve the clinical part of the Tübingen center with the gene therapy treatment of BCM.

The grant we sent will pay the salary of a young scientist and this is a very important aspect of the project because we would like to build a strong new generation of scientists passionate about Blue Cone Monochromacy in particular and inherited retinal diseases more in general.

BCM patients living in Europe would be welcome to get in contact with Dr. Wissinger in the future months. You can contact Dr. Bernd Wissinger at:

Dr. Bernd Wissinger

Bernd.wissinger@uni-tuebingen.de

Web: <http://www.eye-tuebingen.de/wissingerlab/projects/blue-cone-monochromacy/>

- Patient Registry update and some questions for you: a new survey

We are working on the International Patient Registry for BCM. The Registry will be created by an expert company that is working with many other patients' organizations and is able to comply with legal issues such as privacy rules in USA and Europe. Secure servers will encrypt data and patients' information will become anonymous. Access to the database will produce only aggregate data

information and each one of us would be able to have access to the statistical data of all the other patients, comparing his data with the aggregated value of all the others. Scientists will have access to the whole worldwide database and this will produce a central database for all the information about our disease.

Information will be joined to DNA samples stored in a Biobank. We will then be able to send all the DNA samples to scientists who can explain the origin of our disease.

We will set up a steering committee to govern the access to the database.

We will shortly be sending you a survey on this topic. Please check your email box and spam box as well!

- First BCMFF International Meeting in Atlanta – final date and place

We are happy to announce that the first International BCMFF meeting will be in 2017.

Atlanta, Georgia (US) is the city chosen to host the event and dates are August 4/6, 2017.

We will have ophthalmologists, geneticists and scientists speaking about diagnosis, treatment and features of BCM.

2016 Donors – #GivingTuesday event and Bill & Melinda Gates contribution to our cause

Thank you for all your donations !
Bill and Melinda Gates matches giving Tuesday donations.

A special thanks to all the people who sent a donation to BCM Families Foundation in 2016!

We want to thank: Orlando, Daniela, Marisa, Elena, Paolo, Ceci, Maria from Canada, Mark, Elizabeth, Nancy, Janice, Sally, Corinne, Jeff and Erica, Tammy, Equifax, Kay and John, Jasmine, Kim.



#GIVINGTUESDAY™

11.29.2016

- Barbara and Renata attended the New York FasterCures conference

Last November our directors, Barbara and Renata attended the New York FasterCures conference in New York.

BCMFF is a participating organization of TRAIN the FasterCures accelerator.

Barbara and Renata participated to roundtables discussions and met representatives of other patients' associations that maintain a Patient Registry. Moreover they met companies that offer services for the creation of patients' registries.

At the conference Barbara and Renata had the opportunity to meet staff from University of Florida and from other organizations and benefited from learning firsthand about the need to speed up the path toward the cure.



- Please tell us about any foundation or company you know that can give a grant to BCMFF

We have several projects we would like to accomplish in 2017: an animal model for C203R mutation, the BCM international patient registry, the BCM biobank, the support to the University of Tübingen in Germany, the creation of a new generation of scientists working on BCM, the clinical studies of Dr. Jacobson in Philadelphia, a meeting of families with BCM in Atlanta in summer 2017, webinars for our community of patients and their relatives.

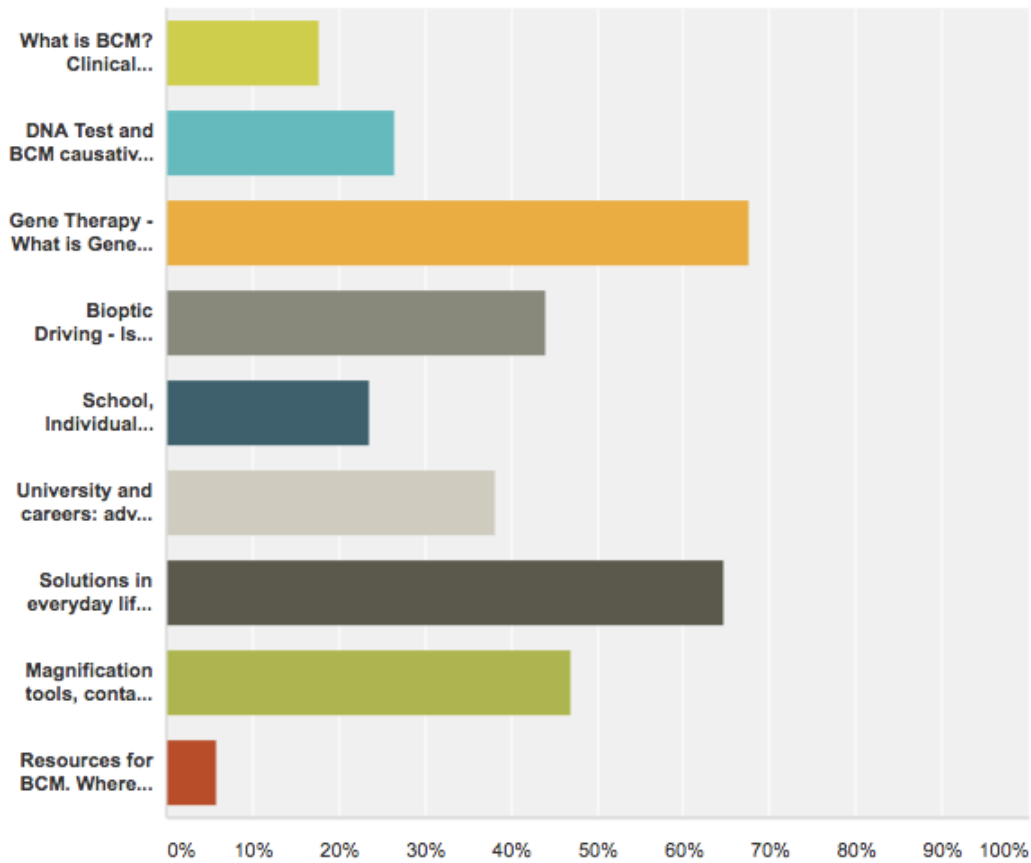
We are looking for sponsors for these projects. Please let us know if you know of any foundations and companies that can give us financial support for these projects.

- Report from our survey about interesting topics

Thanks for answering our latest survey! Here the results:

Which of the following topics would you like us to talk about?

Hanno risposto: 34 Hanno saltato la domanda: 0



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Any Help from you is really appreciated!

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Thank
you!

A warm Thank You!

On our websites you will find there are many ways for donating to BCMFF

Those who donate from US can send a check to:

**BCM Families Foundation
PO Box 7711
Jupiter, FL 33458-7711 USA.**

or make a bank transfer using the following bank details. Please check in advance the amount of fees to be paid for international transfers. For small donations (less than 500\$) Paypal or credit cards are more suitable

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ACCOUNT NAME: BCM FAMILIES FOUNDATION
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Get started





Our President Renata Sarno and the team of BCMFF wishes you all Happy Holidays!
Thanks for your support!



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Toward the cure of Blue Cone Monochromacy

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