



*BCM Families Foundation is registered with the IRS as a 501(c)(3) Public Charity and 100% of donations is going to fund research.*



## **BCM Families Foundation Newsletter n.2**



**Dear BCM Families,**

*Here is the latest news about BCM Families Foundation, people and projects.*

*First of all we'd like to recall for you, or to explain to new families, about **BCM Families Foundation**.*



BCM Families Foundation is a non-profit 501(c)3 organization created in May 2014 by families affected by BCM. Our Board of Directors, working on a volunteer basis, is composed by relatives of people affected by BCM, who have children and grandchildren affected. This is your foundation, where you can serve our common mission: to eradicate BCM by funding the most promising biomedical research that can ultimately reach a cure. Anyone of you can ask to organize a project under the name of BCMFF, can propose projects and activities and can ask to serve on our board, adding her/his skills to ours.

Before May 2014, our President, Renata Sarno, financed privately many research projects since 2010, with the aim to start new research about BCM that could develop a cure.

Renata's sons are affected by BCM and she decided to give back a part of what she has gained in her life in order to help them and other people affected by the same disease. Our history is composed of people, like Renata, Kay, Barb and many of you who helped us, Mark and Diane, Orlando, Paolo, Trudi, Nancy, Corinne, Mari and many others. Many of you participated in our projects and activities.

We are now number more than 180 patients receiving this newsletter. Recently families joined from Italy (Milan), from Florida (2 new families), from Sweden and from California.

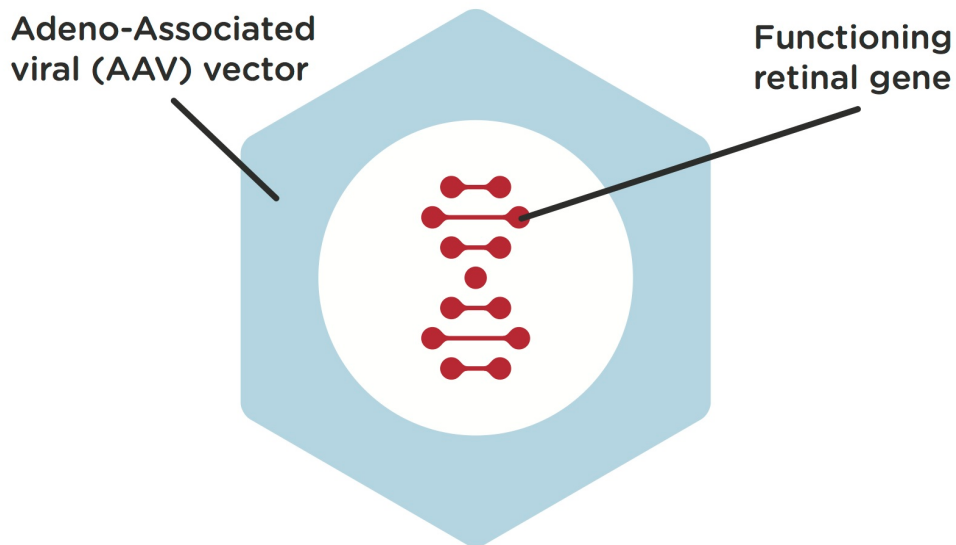
Moreover, a significant piece of our history is BCM FF's support for research projects. These have arrived at important new results, not before available.

Renata financed in 2010 a research project at University of Florida, where Dr. Hauswirth has been working on AAV-based Gene Therapies for eye diseases. With him we developed for the first time an animal model of BCM, and we tested the AAV Gene Therapy on it. This was an exciting new result that has the potential to change the life of many people affected. No one had ever done this before! Restoring visual functions in the animal model is a positive first step. Dr. Hauswirth has been working since 2010 in order to find the best AAV vector for BCM. Dr. Hauswirth is involved in the clinical trial for Achromatopsia. He was elected Florida Scientist of the year in 2009 for his important researches on AAV based gene therapies.

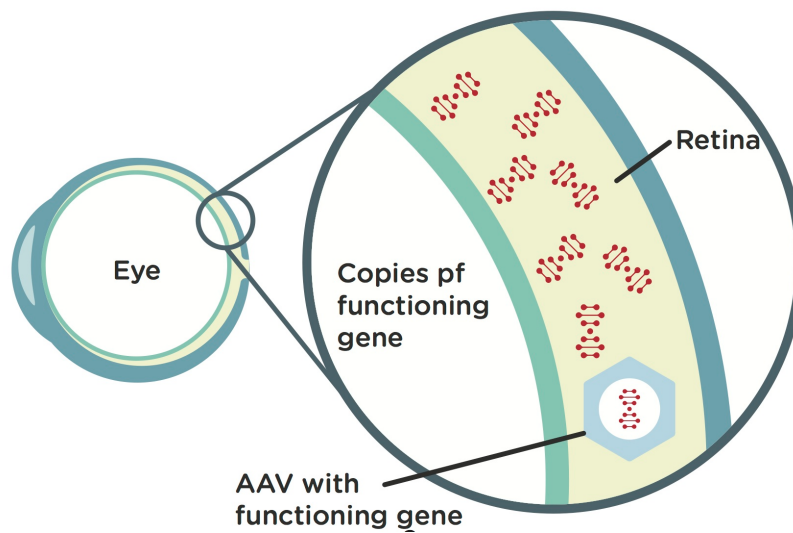
We then started a clinical project at University of Pennsylvania with one of the greatest worldwide experts of inherited eye diseases, Dr. Sam Jacobson. He is performing clinical tests on patients affected by BCM in order to understand if there are enough cone cells in their retinas. These cone cells will be the ones receiving the new opsin proteins with the gene therapy.

The AAV-vector will be carrying opsin protein. The AAV-vector will be injected into a BCM cone cell without opsin proteins, in order to deliver a correction:

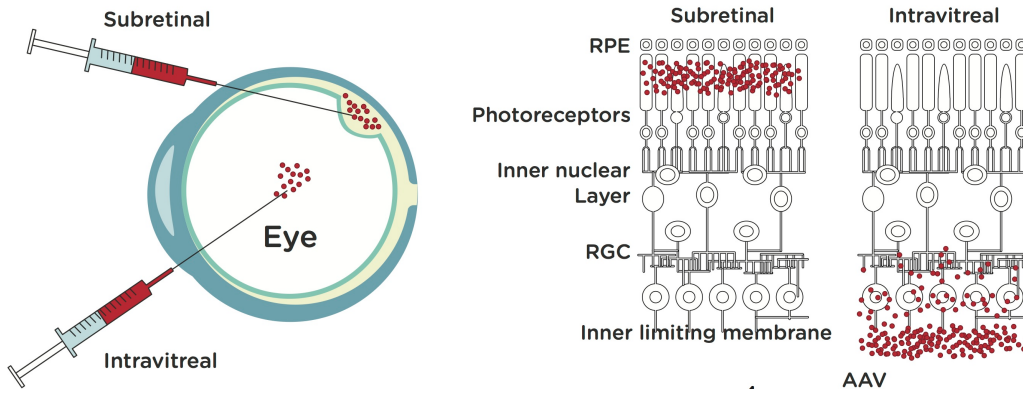
Step 1: Scientists modify an adeno-associated virus (AAV) —a common virus that is not known to cause any disease in humans—by stripping out what's on the inside of the virus and replacing that with a functioning copy of a L-opsin or M-opsin gene.



Step 2: when doctors inject the modified (AAV) vector— in a patient’s eye, it travels into the patient’s retinal cells. Once inside a cell, the vector passes on the functioning copy of the L-opsin or M-opsin gene the patient needs, which was harbored in the AAV vector.



The injection can be subretinal or intravitreal:



Moreover Dr. Jacobson is working on other aspects of the clinical trial for BCM gene therapy: inclusion/exclusion criteria for participants and outcome measures. The question here is: after the gene therapy, what will be the result and how we can measure it ? A scientific approach needs to select measures that can be repeated and that will give numerical results. Result after the cure needs to be better than before and needs to be tested scientifically. Your support and participation to this project has been and is superb.

Then we have a longstanding collaboration with the University of Tuebingen in Germany. We collaborate with Dr. Bernd Wissinger and Dr. Susanne Kohl. Dr. Kohl discovered in the past all the causative genes for Achromatopsia and the Wissinger lab is able to find all the causative mutations of BCM, including the rarest. Our collaboration with them allows our families to have the DNA tested for free, shipping their samples to Germany for the test. At the University of Tuebingen the first humans affected by Achromatopsia CNGA3 have been treated in the past months with AAV-based Gene Therapy. For these reasons University of Tuebingen could become the European center for eye gene therapies. BCM patients living in Europe can consider to be contact with Dr. Wissinger in the next months. You can contact Dr. Bernd Wissinger at:

Dr. Bernd Wissinger

[Bernd.wissinger@uni-tuebingen.de](mailto:Bernd.wissinger@uni-tuebingen.de)

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

*Projects we are financing and we are going to finance are typically discussed with a Scientific Advisory Board (SAB). In our SAB there are recognized scientists serving our mission. Among them, Dr. Jeremy Nathans, who discovered the BCM causative mutations and mechanisms and Dr. John Flannery, very expert on AAV vectors. With them and with you together we can continue to achieve a roadmap toward the cure of BCM.*

*Nearly \$ 2 millions have been delivered up to now directly to scientists, at first privately by Dr. Sarno and then since 2014 by BCM FF.*

*These are the grants that BCM FF has sent since 2014:*

*\$ 147,781 University of Florida - May 2015*

*\$ 25,237 University of Florida - July 2015*

*\$ 102,219 University of Pennsylvania - February 2015*

\$ 99,762 University of Pennsylvania - December 30, 2015

\$ 25,237 University of Florida - January 2016

\$ 99,762 University of Pennsylvania - March 2016

\$ 100,000 University of Pennsylvania - July 2016

*The next grants BCM FF plans to send are:*

\$ 100,000 University of Pennsylvania - February 2017

\$ 38,000 University of Tuebingen - November 2016



Finally, after one year and a half from the first contact, on December 2015 BCM FF reached an agreement with a biotech, AGTC, and the AGTC's commitment to finance the whole BCM Gene Therapy research in the next years. We will discuss with them aspects as final price of the genetic drug and outcome measures. BCM FF would like to represent your voice.

A Patient Registry for BCM is one of BCM FF's significant commitments, we need your collaboration with this project and we will give you more details in the future Newsletter.

Upcoming projects are

- 1) Meeting of families with BCM in 2017, as soon as we will receive final results from University of Florida and AGTC about the AAV vector for BCM.
- 2) Webinars on significant topics of interest to our BCM community.



**BCM and TRAIN**

As you know we are now a participating organization of The Research Acceleration and Innovation Network (TRAIN), a *FasterCures* program established to create opportunities for medical research innovators to discuss and tackle the challenges that cut across diseases. TRAIN is a unique network of patient-driven nonprofit foundations that fund medical research across a spectrum of diseases. They represent the kind of organizations that are fast becoming the engine behind innovation in disease research - collaborative, mission-driven, strategic in their allocation of resources, and results-oriented.

[train.fastercures.org](http://train.fastercures.org)



Any help from you is really appreciated !

**Donate!**

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

**KEY BANK**  
**CLEVELAND, OH**  
**ACCOUNT NUMBER: 776611003867**  
**ACCOUNT NAME: BCM FAMILIES FOUNDATION**  
**ABA: 041001039**  
**SWIFT CODE: KEYBUS33**

International donors can use major credit card on our web site <http://www.blueconemonochromacy.org/donate/>

International wire transfers:

**KEY BANK**  
**CLEVELAND, OH**  
**ACCOUNT NUMBER: 776611003867**  
**ACCOUNT NAME: BCM FAMILIES FOUNDATION**  
**ABA: 041001039**  
**SWIFT CODE: KEYBUS33**

Send us an email at [info@bcmfamilies.org](mailto:info@bcmfamilies.org) with your personal information, your name, full address and email address. We will be able to send you a donation receipt.

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A special thanks to all the people who sent a donation to BCM Families Foundation, especially to Mark and Diane and thanks to volunteers as Nancy, Trudi, Mari and many others.

#### **Help with Fundraising**

Please inform us of any possibility to receive a grant from other non-profit organization, private or public foundation, companies, churches, others. We need help in writing grants proposal and in looking for opportunities to be financed.

We have many projects we would like to finance, first of all the gene therapy project and then our meeting in 2017, webinars and other educational projects.

Please if you have ideas to share, write us and send them to us [info@bcmfamilies.org](mailto:info@bcmfamilies.org)

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By shopping at Amazon.com you will donate a percentage of your purchasing to BCM Families Foundation, while shopping for your favourite items.

Bookmark this link so your shopping will benefit your favourite charity: <https://smile.amazon.com/ch/47-1327738>

Amazon selected BCM FF for a test of a few weeks, monitoring how many people have chosen our charity and what kind of products they buy.



*Our President Renata Sarno and the team of BCM FF wishes you all a beautiful summer!*

*Thanks for your support!*



**BCM  
FAMILIES  
FOUNDATION**

Toward the cure of Blue Cone Monochromacy  
PO Box 7711 - Jupiter FL 33458-7711 USA

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