BCM FAMILIES FOUNDATION
Toward the cure of Blue Cone Monochromacy
PO Box 7711 - Jupiter FL 33468-7711 USA

BCMFF Spring 2018 Newsletter

Dear **BCM Families**,

A lot has happened so far and we would like to share this news with you.

We are hoping for your continuous support, in order to be able to advance the research, as we have done so far.
We want to warmly thank you for donations received during the last **#GivingTuesday.** The **$14,202.55** received has been used to support the **animal model** project, that you will find discussed in this newsletter.
This extraordinary result is due to the effort of all fundraisers and to a special organizing Subcommittee - Kay McCrary, Rachel Workman & Nancy Noel Lemon - and is an image of the light that is in this community of people who are walking together towards a **#Cure4BCM**.

A special Thank You to Corinne Harmon's family, because generous donations in memory of her father, Lewis Bohannon, helped us so much.
Thanks to the donors who kindly supported our mission with their donations in recent times: Daniel and Mary Beth O'Donnell, Debby Taylor, Janice and David Ishee, the Lambert family, Lorna Finch.

We want to remind you that **20th of May** is the **International BCM Awareness Day**. Any initiative toward the organization of an event is welcomed! You will find details on how to support our mission by engaging actively, below in the newsletter.

Thank you all!
BCMFF

In memory of Lewis Bohannon

Lewis was one of a kind…overcoming every obstacle he faced. His condition never stopped him and he was a successful entrepreneurand remarkable husband, father, and grandfather. A man of integrity, generosity, and faith.

Thank you Lewis

We need your feedback!

Please help us by answering this **one-question survey**.
Please only answer one question:
1)If you are a person affected by BCM, you can answer the first question.
2)If you are a parent/carer of a BCM under-18, you may answer the question relating to minors

Thank you!

1. For BCM Patients
How often do you visit your Ophtalmologist/Clinician?

|SURVEY: Once a year|
|SURVEY: More than once a year|
|SURVEY: Once every two years|
|SURVEY: Once every five years|
|SURVEY: Less than once every five years|

2. For parents or guardian of a BCM under-18 patient
How often do you bring your children to their Ophtalmologist/Clinician?

|SURVEY: Once a year|

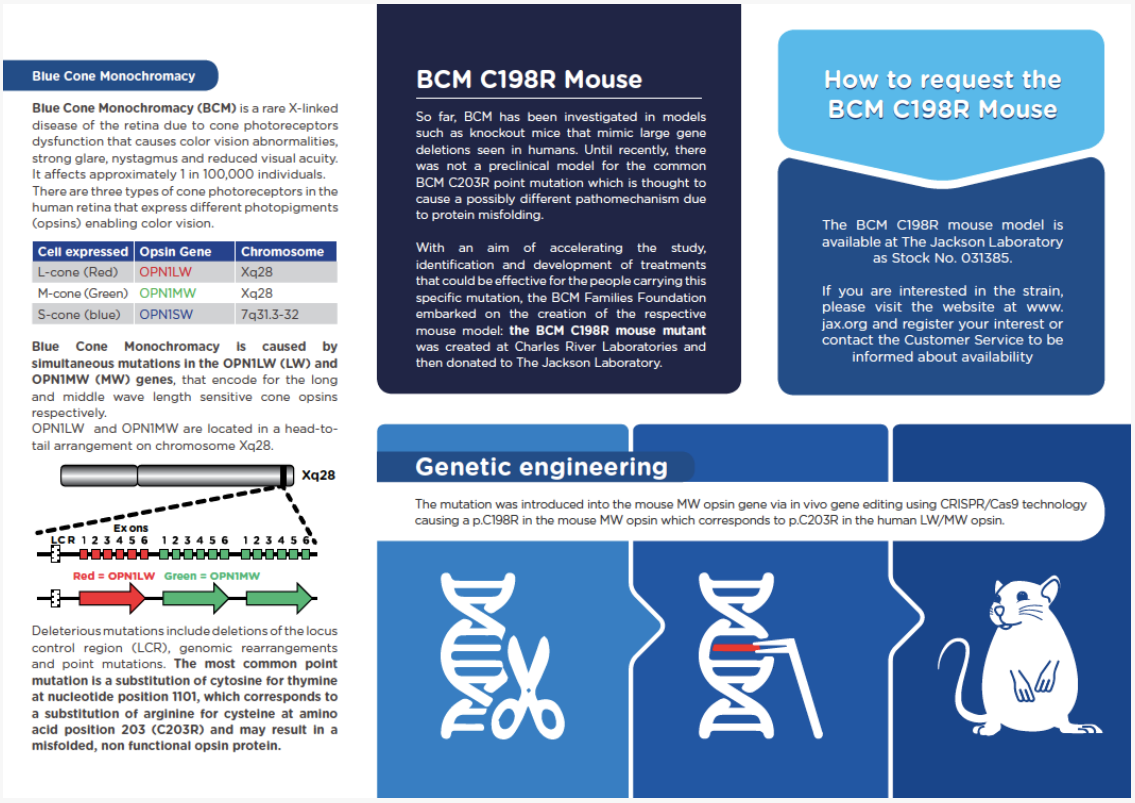
|SURVEY: More than once a year|

|SURVEY: Once every two years|

|SURVEY: Once every five years|

|SURVEY: Less than once every five years|

BCM Animal Model



BCMFF obtained an extraordinary result: the creation of a new experimental model, the BCM C198R mouse. This mouse line was created with CRISPR/Cas9 technology, to precisely model the cysteine-203-to-arginine (C203R) Blue Cone Monochromacy mutation found in the human LWS/MWS cone opsin gene(s).

This amino acid substitution mutation is responsible for Blue Cone Monochromacy in a large percentage of all people affected by the disease world-wide. The BCM C198R mouse line is the first preclinical model available for studying the specific pathomechanisms associated with Blue Cone Monochromacy and to test potential therapeutic approaches.

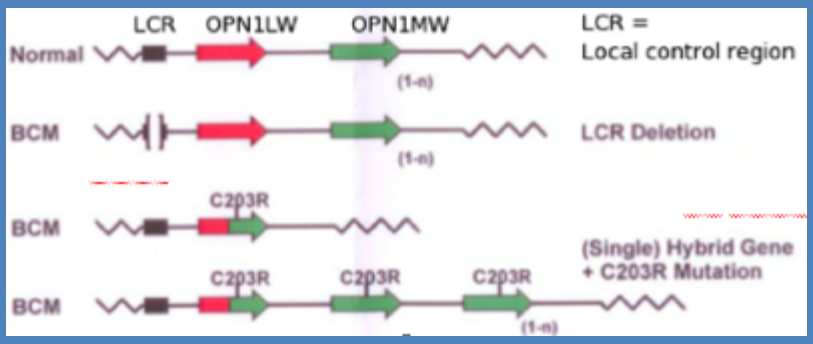
The model was established by the BCM Families Foundation (BCMFF) with the

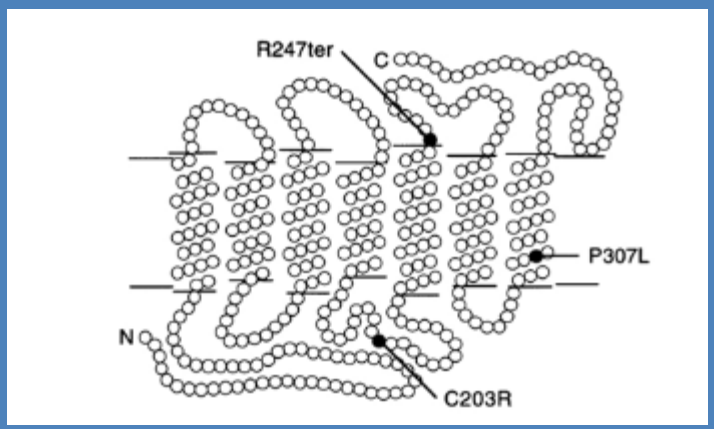
ultimate aim to accelerate the pace of research towards clinical translation of potential therapeutic interventions.

Towards this aim, BCMFF contracted for the creation of the mouse model with Charles River and then donated the strain to The Jackson Laboratory where interested research institutions, as well as biotech/pharma companies, can obtain it.

Now that this mouse model is publicly available, we hope to gather the interest of outstanding scientists and researchers.
The model has already been delivered to University of Florida and University of Tubingen.

We are now asking for your help and donations; we need $25.000
in order to start research projects on this mutation and, therefore, continue to walking our way toward the cure of BCM.





C203R Point Mutation

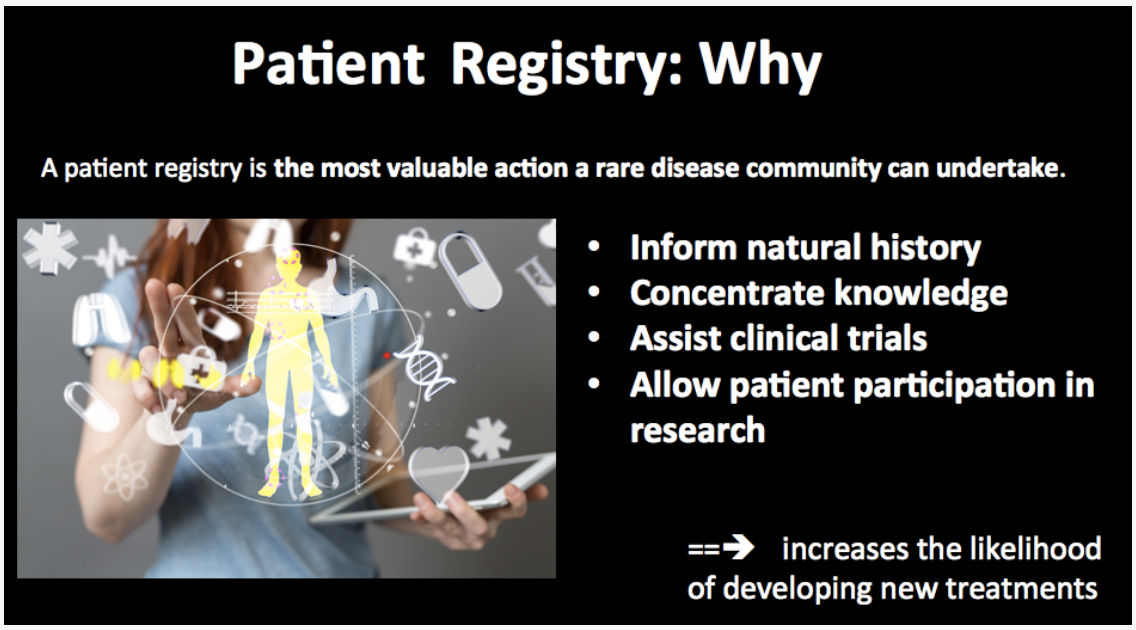
BCM International Patient Registry

BCMFF has been working on the creation of an International Patient Registry.
We are still working on this project, which is taking shape, together with our software company partner, Open App.

BCM Registry will collect and store personal and clinical data of patients with BCM: its primary objective will be to gather as many families/patients as possible with proven BCM, in order to allow further contacts with them and involve them into future clinical trials.
Provided that the data stored in the registry belong to the patients who consent to share their data for the purposes described above, the Registry platform is being set up by BCMFF that will keep control and responsibility over it.

BCM Registry will be a patient-powered registry as patients will register themselves and provide basic information (demographic and basic clinical data such as diagnosis and symptoms). Therefore, the registration will be as easy as possible and the software interface will be set up in order to be BCM-friendly.

More importantly, the project will be compliant with the new European regulation regarding the privacy of patients' data, specifically sensitive data.



The starting of the work on the registry was made possible by the support received by Waldesian Church, however we need your help to advance with this project. We hope to collect $20,000.

Stay tuned! More information on how to participate to the Registry will come!

We are proud to post the result of a study by the University of Florida, made possible by the support of BCM Families Foundation, on gene therapy testing in mice.

Gene Therapy improves daylight vision, color vision deficiencies in animal model

By: Doug Bennett, University of Florida

For people with blue cone monochromacy, the world is blurry, colorless and uncomfortably bright. Now, University of Florida Health researchers have found a gene therapy that restored crucial visual functions to affected cone photoreceptor cells during testing in mice.

While additional study is needed, researchers say the findings are proof of concept that the gene therapy works in an animal model and should be considered for a human clinical trial. The findings were published recently in the journal Molecular Vision.

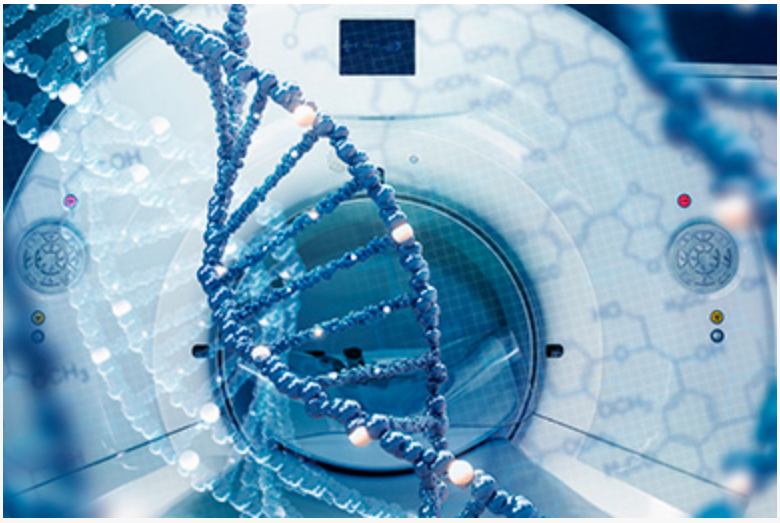
Read More

Wen-Tao Deng, Ph.D., is an assistant scientist in the UF College of Medicine's department of ophthalmology research.

AGTC Latest News

The collaboration with BCMFF and AGTC is going on and we are happy to announce two important projects on which AGTC is focused: **Retinitis Pigmentosa** and **Retinoschisis.**

For more information on AGTC and its pipeline of AAV-based gene therapy candidates in rare disease, please visit www.agtc.com/programs/



AGTC Doses First Patient in Phase 1/2 Clinical Study of Gene Therapy for the Treatment of X-Linked Retinitis Pigmentosa

April 18, 2018
GAINESVILLE, Fla., and CAMBRIDGE, Mass., April 18, 2018 (GLOBE NEWSWIRE)

Applied Genetic Technologies Corporation (NASDAQ:AGTC), a biotechnology company conducting human clinical trials of adeno-associated virus (AAV)-based gene therapies for the treatment of rare diseases, today announced that it has dosed the first patient in the Company's Phase 1/2 clinical trial evaluating the safety and efficacy of an investigational AAV-based gene therapy for the treatment of X-linked retinitis pigmentosa (XLRP). The multicenter study will assess AGTC's novel recombinant AAV vector expressing a human RPGR gene (the rAAV2tYF-GRK1-RPGR) in patients with XLRP. The patient is being followed by Dr. David Birch of Retina Foundation of the Southwest and the surgery was performed by Dr. Andreas Lauer of Oregon Health & Science University.

Read more

AGTC Announces Completion of Enrollment of Phase 1 / 2 Clinical Study of Investigational Gene Therapy in Patients with X-linked Retinoschisis (XLRS)

April 10, 2018
GAINESVILLE, Fla.

CAMBRIDGE, Mass., April 10, 2018 (GLOBE NEWSWIRE)

Applied Genetic Technologies Corporation (NASDAQ:AGTC), a biotechnology company conducting human clinical trials of adeno-associated virus (AAV)-based gene therapies for the treatment of rare diseases, today announced the completion of enrollment in a clinical study of the company's gene therapy product candidate, in collaboration with Biogen, for the treatment of xlinked retinoschisis (XLRS). This multicenter study is designed to evaluate the collaboration’s AAV vector expressing retinoschisin (rAAV2tYF-CB-hRS1) in patients with XLRS caused by mutations in the RS1 gene. Topline data are anticipated by Q4 2018 with the final analysis at the twelve-month time point.

Read more

It is time to take action!

BCMFF would like to ask you to join us as ambassadors of BCMFF, and in doing so, organizing your own special event, inviting everyone you know to learn more about BCM!

We are pleased to announce that we are focusing on growing our amazing community and increasing our efforts in the US to spread awareness and raise funds for scientific research. As part of this endeavor, we are introducing the BCMFF Community Engagement Initiative to give those interested an opportunity to become BCMFF ambassadors and become more involved with the foundation.

In order to continue to grow our base of support and to build a larger understanding surrounding this disease, it's important that we continue to educate the public and raise awareness.

For those of you looking to represent a cause close to your heart and become an ambassador of the BCM Families Foundation, we have developed the tools to guide you. Within, you will find a toolkit to aid in the planning and execution of your own fundraiser through a step-by-step guide and helpful customizable templates.

Download the guide at www.blueconemonochromacy.org/community-toolkit/ and start organizing an event

Please feel free to reach out to John Cavitt, who is coordinating all activities

(john.cavitt@bcmfamilies.org). The support BCMFF receives from our community helps ensure our mission to empower patients and lead a path towards a cure for BCM.

Thank you for your support,
BCMFF

60 Seconds with...

by Trudi Dawson

Name: Virgil E Wade

Age: 83

Where do you live? Lakewood, OH

How many relatives do you have with BCM? 5 living

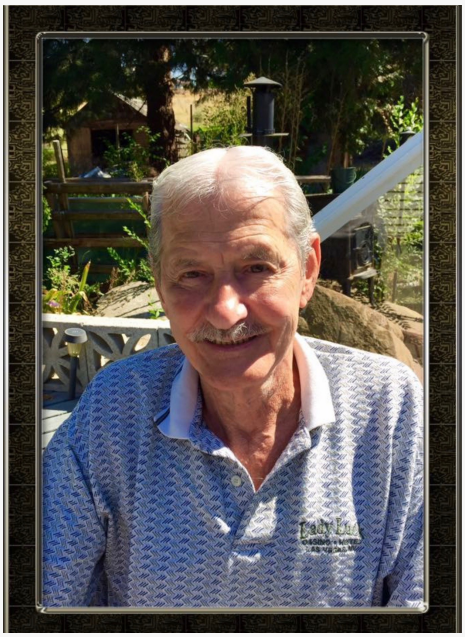
What is your job? I was a Machinest. I would have liked to have been a Cowboy and Rancher all my life

What are your hobbies? Playing the guitar, writing music and songs, and horses

What is your most useful BCM tip? Always carry sunglasses with you

Greatest achievement/proudest moment so far: Marrying my wife, the mother of my 3 daughters and making a life for them.

Not many people know this about me but... I was a horse trainer for Adele Ohl Van Parker who was the most famous horsewoman in the world at Parker Ranch in North Olmsted, OH. She cared for horses of famous stars like: Gene Autry and performed with Wild Bill Hickok. She trained horses to jump off diving boards. I cared for her favorite horse Traveler.



Our mailing address is:
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