



**BCM  
FAMILIES  
FOUNDATION**

Toward the cure of Blue Cone Monochromacy

PO Box 7711 - Jupiter FL 33468-7711 USA

# HAPPY EASTER!

Dear Families, Dear Friends,

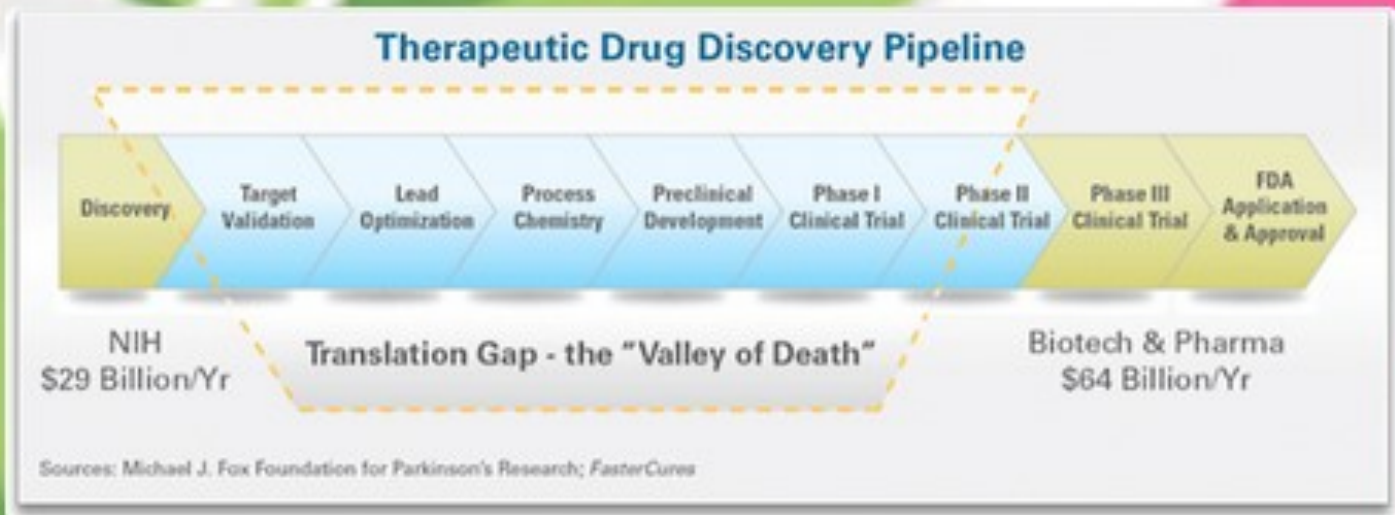
This year the wishes for a happy Easter find us all in an unprecedented situation, which puts us to the test, fueling fears and uncertainties. We know that the whole world is united in the fight against the SARS-Cov2/Covid-19 virus and our hope is that, in these days of forced lockdown, the importance of the influences and the presence of others can be rediscovered, even if at a distance.

The world at the end of this epidemic will be different from the world we left behind, of which we now see strength and weakness, beautiful aspects that we miss and ugly that we would like to let go of. Easter is a feast dedicated to re-birth and we hope that everyone can have a dream on this day to rebuild a more welcoming, healthy and beautiful world at the end of this outbreak.

Our dream is to land in a world in which there will be a clinical trial for BCM gene therapy, and we will talk about this in the first article.

BCM Families Foundation

## What is a Drug Pipeline?



Although Covid-19 is an infection that has forced half the global population to lockdown, the road to reaching a vaccine is long and the forecast for obtaining the final drug on the market is around 1 year or more. Some pharmaceutical companies are starting the road that will take them to the clinical trials of the vaccine.

What is it that takes so long to get a great new idea into the clinic to benefit patients sooner and faster than that?

What do we learn from Covid-19 that will be useful for gene therapy for BCM?

When you open a site dedicated to the Covid-19 vaccine for example:

<https://www.hhs.gov/about/news/2020/03/30/hhs-accelerates-clinical-trials-prepares-manufacturing-covid-19-vaccines.html>

You find words like pre-clinical results, sponsor, regulatory process, IND submission to FDA, Phase I, II or III of a clinical trial, manufacturing and production. Let's see how the process takes place, because for Blue Cone Monochromacy, we are going the same way... [read more](#)



# WARNING

If you are hospitalized with Covid-19 symptoms please inform doctors that you are suffering from BCM. In fact use of chloroquine and hydroxychloroquine **can cause retinal toxicity** that mimics inherited retinal diseases. Be aware that a patient with BCM taking these drugs for COVID-19 would be very difficult to monitor for toxicity of the drug versus BCM effects.

"We do not know if these drugs can **accelerate** the visual loss in BCM, for example – that is unknown and would be a big concern. We may know some formula about how long and how much it takes to intoxicate the retina in those with normal vision but we have no idea what dose or how long it would take to worsen BCM retina."

Prof. S.G. Jacobson – Scheie Eye Institute – University of Pennsylvania

"The risk of irreversible maculopathy at these higher doses for short periods of time is unknown. Patients should be informed of the potential for macular toxicity before starting therapy." [American Academy of Ophthalmology](#).





## **SARS-CoV2 Coronavirus and Gene Therapy Adeno-Associate Virus**

Blue Cone Monochromacy Gene Therapy is based on Adeno-Associated Viruses (AAV) that will be modified to carry Opsin proteins inside the human retina and injected intravitreally in the eye...[read more](#).

*"AAV compared to the COVID19 Coronavirus, it is a simple comparison: 1) AAV and COVID19 are in no way biologically related to each other. And most importantly, 2) AAV currently infects more than 80% of the world's population but causes no disease after 40 years of study. In stark contrast, after just a few months COVID19 has infected a small but growing fraction of many nations of the world and causes a terrible, often fatal, disease".*

Prof. W.W. Hauswirth – University of Florida

## BCM Registry – Statistics

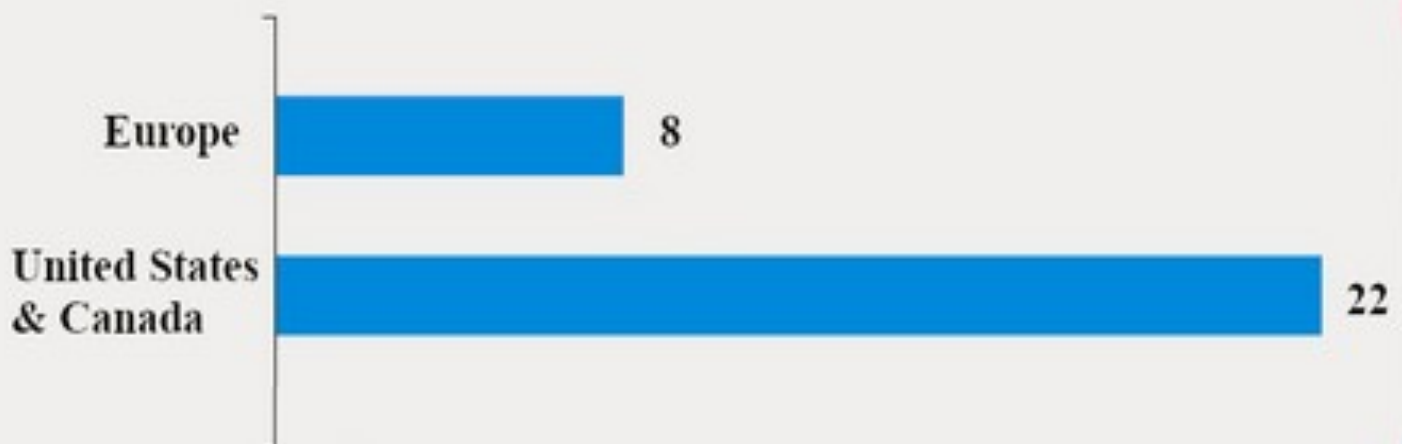
We are proud to share with you the first statistical data about the BCM Patient Registry.

The Total Number of enrolled and validated patients is 30. We have more than 25 additional people who are waiting for their DNA test result or their clinician, to enroll.

The BCM Registry includes more United States and Canada residents than Europeans. So, Europeans, please: hurry up!

We have more patients with deletion as causative mutation than C203R point mutation. However, 7 people are still waiting on their clinician who has to input genetic mutation. Please call your doctor and push her/him to insert your causative mutation!

Statistic Patient Validated by Country





## Statistic Patient Validated by Mutations



### Genotype Mutations:

N/A = "Lack Diagnosis"

DEL = "Gross Deletion"

REA = "Other Structural Rearrangement"

PT1 = "Point Mutation C203R"

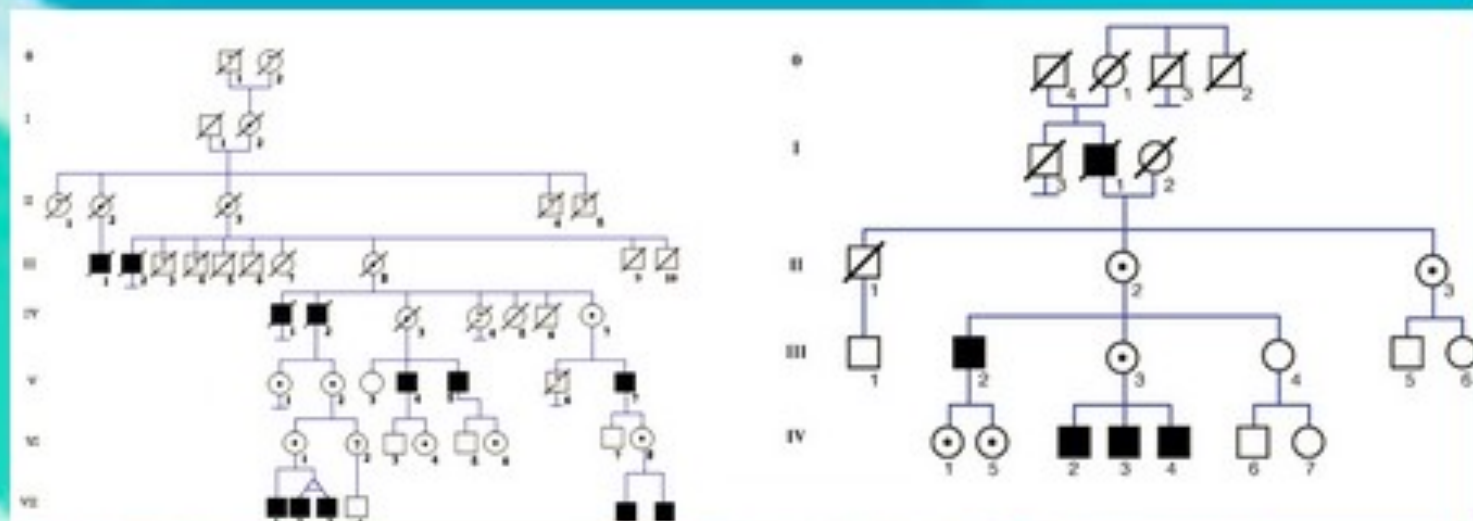
PT2 = "Other Point Mutation or small Ins./Del."

EX3 = "Exon 3 splicing deficient haplotype"

OTH = "Other"

\* zero values are not displayed.

Finally, in the Registry there are a total of 9 families and 8 beautiful Pedigrees. Please contact our Registry Manager at [Registry.Manager@BCMFamilies.org](mailto:Registry.Manager@BCMFamilies.org) if you want to draw your family pedigree!



**Join the Registry today!**

## BCM Patient Registry Testimonials

Here you find some words and thoughts regarding our BCM Patient Registry and what it means, written by patients who have enrolled so far.

"Dear BCM family,  
the virus will go – but BCM will stay. BCM is not life-threatening, but those affected have to struggle with the limitations throughout their lives.

Surely at some point you have come to accept the limitations. Do we want to make the difficult way until then also for our children?

Should we not do everything in our power to prevent our children from doing so? That is exactly the goal we are working towards together.

This is the goal that BCMFF is working towards tirelessly. At the end there is a healing possibility.

Exactly at this point the current crisis and our BCM concern is the same. The goal is the healing.

What does it take to reach the goal as quickly as possible? Data from tests – as much data as possible.

The results of the tests must be available for all those working on the cure. This is exactly what our patient registry at BCM is for.

The better the information is and the more comparative data is available, the better conclusions can be drawn.

Which ultimately increases the chances of a cure.

Talking to family members. It will not always be successful. But anyone who registers and submits their DNA test results will take us one step closer to our goal.

I wish everyone, despite the current problems, a wonderful Easter.

The spirit of Easter should give hope to all. Stay healthy, everyone. Best wishes"

Hawkmaster



We want to warmly thank our BCMFF volunteer, Simona, who collected \$340 for the BCMFF by selling guided tours of the city of Rome, Italy, with the touristic guides of [RomAspasso Association](#).

Thank you from the BCMFF Community!



## Coming Summer 2019, Blue Miracle Coffee Company



This is a coffee company that has a dual purpose of sending proceeds to BCM Family Foundation and helping the impoverished in Nicaragua. Shepard Cavitt, 12, with BCM wanted to design a product that could be a business for him, help towards our cure, and help out some new friends in Nicaragua. Please be on the lookout for the launch this summer at [bluemiracle.coffee](http://bluemiracle.coffee) for products including k-cups and others. These will be distributed on Amazon, some stores, and all products will be available at cost to any BCM family for their own fundraising efforts. You can email the Cavitt's at [thecure@bluemiracle.coffee](mailto:thecure@bluemiracle.coffee).

## 60 Seconds with...

By Trudi Dawson





**Name:**

Jameson Cavitt

**How old are you?**

8 years old

**Where do you live?**

Sugar Land, Texas USA

**How many relatives do you have with BCM?**

2 brothers, 1 grandpa and 3 cousins all have BCM

**What is your dream job?**

When I am grown up I would like to study animals and help them to survive if they are hurt.

**What are your hobbies?**

Playing with my pet Bird Zoë, swimming and drawing are some of my favorite hobbies. Also baking with my mom is fun! I have two dogs, a frog, a snail, fish, shrimp, and a bird.

**Useful BCM tip?**

Not being afraid to ask for help from friends ♥

**What is your biggest achievement so far?**

I am most proud of being able to make Level 5 in gymnastics.

**Not many people know this about me but...**

I am an excellent artist and I love to design and name roller coasters!

BCMFF needs your **support!**  
Today you can make the difference:

**Donate now!**

