

## Summer Newsletter 2024

BCM Families Foundation <info@bcmfamilies.org>

Wed 7/3/2024 5:02 PM

To:info@bcmfamilies.org <info@bcmfamilies.org>





Dear Friends, Families, Ambassadors, Volunteers

Happy Summer and Happy Holidays!

With this letter I update you on the status of BCMFF projects to ask you to help the BCMFF with fundraising activities.

Select your preferred project and help us raising funds:

**Project #1.**

University of Pennsylvania - Dr. Cideciyan and Dr. Aleman

The aim of this clinical project is to define endpoints of a gene therapy clinical trial and to write the clinical protocol for the trial + a description of all outcome measures of the trial. Moreover, UPenn is interacting with another clinical center, Tübingen in Germany, to share knowledge about measures and verify reproducibility of them. Outcome Measures are the result of 14 years of clinical projects done by Dr. Samuel Jacobson with the support of the BCMFF.

Total of \$148,759 initially granted + \$35,000 Extension

To be delivered: \$49,587 + \$35,000

**Deliverables:**

- 1) A study report detailing clinical findings regarding potential outcome measures for a gene therapy treatment of BCM, for example ADVN-062.
- 2) A written document that can be used for a future clinical trial, i.e. a clinical trial protocol with descriptions of Standard Operating Procedures for performing key outcome measures.

Last payment made on June 2024: \$17,500

**Total to be paid in 2024: \$66,087**



University of Pennsylvania Clinical Studies project is supported in 2024 by funds of Konecta Italia S.p.A received through Associazione Acromati Italiani onlus.

**Project #2.**

Tübingen Clinical Center – Dr. Katarina Stingl

The aim of the project is to buy an instrument for the Tübingen clinic, to allow the principal investigator, Dr. Katarina Stingl, to test outcome measures as planned by UPenn in her clinical lab. The instrument is the Metrovision Static Perimeter.



Total needed amount Euro 52,000 = \$56,500

BCMFF President will gift \$35,000

**To be delivered in 2024: \$21,500**

A fundraising campaign has started in Italy where people can donate 5 Euros every 1,000 Euros of their taxes for non-profit projects. Learn more on 5x1000 campaign. <https://www.blueconemonochromacy.com/events/5x1000-fundraising-campaign-2024/>

### **Project #3.**

No-Cost DNA Test program for Blue Cone Monochromacy with the Wissinger Lab at the Tübingen University.

The Wissinger Lab is the only laboratory in the world able to perform DNA test on BCM for all causative mutations. Thanks to the BCMFF President and to the BCMFF, since 2009, the No-Cost DNA Test program for BCM is active, allowing all members of our community to be tested with no cost for the test. Several doctors every year send to the Wissinger Lab samples to be tested for BCM too. A total of around 55 tests per year are performed on male patients with a clinical diagnosis of BCM and females, older than 18, that want to certify their status of carriers.

Total of USD 27,300 per year – **Fully financed for 2024!**



No-Cost genetic diagnosis project is supported in 2024 by funds of [Konecta Italia S.p.A](#) received through [Associazione Acromati Italiani onlus](#).

Thanks to restricted donations received from some of you and a large donation from Pratico Family, BCMFF has been able to send \$16,500 and to fully finance the program for 2024. So, for 2024 our community and all doctors who have patients with a clinical diagnosis of BCM can benefit from a No-Cost DNA test program. If you need to perform the DNA test, please hurry up and do it in 2024!

### **Project #4.**

Collaboration with the Institute of Molecular and Clinical Ophthalmology Basel (IOB) – Basel, Switzerland alongside the University of Tübingen for improving DNA Tests for Blue Cone Monochromacy and inserting them into Next Generation Sequencing automated tests.

IOB software could find the likely genetic cause in a big fraction of cases.

Total Euro 8,070 = \$ 8,700 – **Fully Financed**

### **Project #5.**

At West Virginia University, Dr. Wen Tao Deng is working on testing AAV vectors for

gene therapy, using animal models developed by BCMFF in collaboration with our Scientific Advisory Board

Total of \$130,888

**To be delivered in 2024: \$30,888**

#### **Project #6.**

Search Engine Optimization SEO improvement and new web site for BCM-- financed for 2024 thanks to our President.

The Website of the BCMFF has been re-organized and designed to attract more people looking for a diagnosis. The work has been done by a staff of people who worked on new web pages, new contents organization, translations, Search engine Optimization, graphics. Please visit the new website at <https://www.blueconemonochromacy.org>.

Particularly visit the new web pages:

<https://www.blueconemonochromacy.org/blueconemonochromacy/>

<https://www.blueconemonochromacy.org/nystagmus/>

We can use Google Adv grants to improve our positions on Google Searches.

**From 2025 it will cost \$12,000 – to be financed.**

#### **Project #7.**

Our [International Patient Registry](https://www.BCMRegistry.org) is active at <https://www.BCMRegistry.org> and there are people working to maintain it online. There is a Registry Manager, a Data Protection Officer, a Software House, an Institutional Review Board, all working to keep it active. Here you have the 2024 Poster with aggregated data. Several projects can be done.

**BCM Patient Registry - \$5,700 for 2024 - Fully financed yet.**

You can support the BCM Projects by donating to the BCMFF and organizing fundraising events.

For more information, visit:

<https://blueconemonochromacy.com/community-toolkit/> - where you can also download our brochures.

If you need our logo, please contact us via email, and we will send it to you digitally.

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#### **In this Newsletter:**

- 1) Dr. Mark Pennesi at the Retina Foundation
- 2) Newly Redesigned Foundation Website
- 3) Sunglasses for Summer Fun by Dean Monthei

4) 60 seconds with... An interview to Mike Fry by Trudi Dawson

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Thank you for helping us in our mission to reach the cure for Blue Cone Monochromacy. Your help is essential and will make a difference!

## Donate

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### **1) Dr. Mark Pennesi at the Retina Foundation**

Dr. Mark Pennesi, expert on Inherited Retinal Diseases, has recently arrived to the [Retina Foundation](#), in Dallas, TX.

Dr. Pennesi is the Director of Ophthalmic Genetics. He will focus on expanding the Retina Foundation's world-class center of excellence in inherited retinal degenerative diseases to bring cutting-edge treatments from the laboratory to the clinic.

*"Very pleased to take 2 our boys to Retina Foundation in Dallas. Its been too long since we had a checkup. Mark Pennesi, MD, PhD and his staff took over 6 hours with us to do a thorough checkup. They offer these services to all IRD patients at no cost. Dr. Pennesi is a leading clinician and researcher in our field and we were honored to sit with him and have him give us a very thorough evaluation. Their staff was very friendly and professional. "*

- John Cavitt



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## 2) Newly Redesigned Foundation Website

We are excited to announce that our foundation's newly redesigned website is now live and can be accessed at [the same address](#).

The updated site features comprehensive and informative pages, including a newly updated section on Blue Cone Monochromacy.

Additionally, we have introduced new pages dedicated to [Blue Cone Monochromacy](#), [Nystagmus](#), [Color blindness](#), [Achromatopsia](#), and [Inherited Retinal Diseases](#).

We invite you to explore these resources and learn more about these topics and our ongoing efforts to support those affected by BCM.

**Check it out**

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## 3) Sunglasses for Summer Fun

by Dean Monthei





The following advice might not apply equally to everyone with BCM since the light sensitivity varies person to person.

I always have at least 2 pairs of sunglasses with me for full sun and cloudy conditions. I have a deep red pair that greatly enhances colors for me in bright sun (great for flowers, clothing and car colors). The lighter pair (NOIR #70) is reddish and does not enhance colors as much but keeps me from squinting (normally a problem for me even on cloudy days). NOIR apparently discontinued the #70 color but any sunglasses will do for cloudy weather. Both of mine are fit-over style so I can use them over glasses. Wraparound sunglasses called FastCap SG-AF-R510 are the same color as the wraparound glasses I use for bright sun (but they break easily).

In some situations where color is not a concern, I have also found a pair of gray sunglasses that adjust how dark they are by rotating a small gear mounted in the middle. They are based on 2 polarizing lenses that when rotated relative to each other (crossed polarizers), they go from cutting out about half the light to extremely dark. These are very useful when I work in my yard switching back and forth from direct sun to working in shadows. I have added rubbery side shields to help block stray side light.

Some people with BCM and Achromatopsia are so light sensitive that the glasses I use are not dark enough for them. NOIR Medical makes glasses that are a very dark red transmitting about 5% of visible light (NOIR color #99). NOIR Medical makes a huge number of frame styles including fit-over, wraparound, goggles style, and wire frame aviators. This includes frames for children (pediatric). They also have a sample lens color "lens ring" with a whole bunch of colors including #99 to see what works for you (\$80). Unfortunately they do not make lenses in the variation of red I find most useful for enhancing colors.

Airports are usually very comfortable for me but when traveling alone having a range of sunglasses is very important to me. Instead of going down a gangway from the terminal to the plane, I occasionally have had to walk out on a tarmac (open asphalt in blinding bright sun). There are also a few extremely bright spots inside terminals like Dallas that I really needed sunglasses. Having a monocular and folding hat in my



pocket is also almost a necessity for me in summer especially when traveling alone.

**Links:**

- Favorite red fit-over sunglasses Laser Safety Glasses Protection Goggles for Green 532nm laser (Pack of 3 for \$13) [https://www.amazon.com/gp/product/B099WHTB7B/ref=ppx\\_yo\\_dt\\_b\\_search\\_asin\\_title?ie=UTF8&psc=1](https://www.amazon.com/gp/product/B099WHTB7B/ref=ppx_yo_dt_b_search_asin_title?ie=UTF8&psc=1)
- FastCap SG-AF-R510 - red wraparound safety glasses - ~\$8.40 <https://www.rokhardware.com/fastcap-cateyes-safety-glasses-red-lens-no-magnification.html>
- Adjustable darkness polarizing glasses - ~\$8.50 <https://www.ebay.com/itm/371845397940>
- NOIR #99 Low Vision red glasses – Price varies a lot with frame style - very dark <https://www.noirinsight.com/catalogsearch/result/?q=%2399>
- Folding hat (cap) [https://www.amazon.com/gp/product/B0CGTYSB8L/ref=ppx\\_yo\\_dt\\_b\\_asin\\_title\\_ooo\\_soo?ie=UTF8&psc=1](https://www.amazon.com/gp/product/B0CGTYSB8L/ref=ppx_yo_dt_b_asin_title_ooo_soo?ie=UTF8&psc=1)
- Side shields for use with adjustable sunglasses [https://www.amazon.com/gp/product/B09WR1Y842/ref=ppx\\_yo\\_dt\\_b\\_search\\_asin\\_title?ie=UTF8&th=1](https://www.amazon.com/gp/product/B09WR1Y842/ref=ppx_yo_dt_b_search_asin_title?ie=UTF8&th=1)
- NOIR Filter Ring <https://www.noirinsight.com/l-ring>



#### 4) 60 seconds with... An interview with Mike Fry

by Trudi Dawson



60 seconds with.....

**Name:** Mike Fry

**Age:** 44

**Where do you live?**

I live in Carmel, Indiana: however I was born and raised mostly in Northern California, which is where I met and married my wife. A little over 10 years ago we moved to the East Coast for her to attend a great business school. She entered a career in finance, and when Covid hit we moved to Indiana, which is more affordable but still a short flight to New York. We visit family in California at least once a year and my wife flies to NYC several times a month.

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

I have at least 6 close relatives with BCM: my younger brother, 3 cousins from my mom's identical twin sister, my mom's cousin, and my mom's grandfather.

**What is your job/would you like your job to be?**

I am a stay at home dad to 5 wonderful children. Before that I was an attorney for the State of California in the Department of Health Care Services.

**What are your hobbies?**

My hobby is exercising. Each day I try to lift weights for about 45 minutes, do an hour of cardio on the elliptical, and walk 10,000 steps.

Another hobby of mine is practicing Catholicism. I pray several times a day, listen to Catholic podcasts ect., regularly attend Sunday mass, and receive the sacraments. I love it. I can not adequately express how much comfort and solace it gives me. As GK Chesterton said "the church is larger on the inside than it is on the outside" - it's an inexhaustible resource.

Another hobby of mine is listening to news, particularly the Wall Street Journal and New York Times. I also very much enjoy scrolling through X (formerly Twitter).

**What is your most useful BCM tip?**

When I was about 25 years old I started using surgery scopes as a low vision aid for reading. They are humongous and embarrassing to wear around people in public, so

I can't recommend them for everyone, but nevertheless they let me read comfortably at home, which has improved my life in some ways.

***What would you tell younger BCM boys/your younger self?***

If you learn and accept that you are visually impaired, it can be a relief because it explains why you can not do so many things others do easily. The psychological baggage that goes with having BCM is extreme. You have excellent mobility so at some level you forget that you're very visually impaired. Remember 2 things: 1) be a good person, do not sin, have a clean conscience- this will relieve some of the anxiety that is inherent from having a major disability: 2) do not be ashamed of having a disability, but accept it because it explains so much of the difficulties in your life. Don't be overly ambitious, perhaps seek a profession in the visually impaired community. Not only will you be helping people more unfortunate than yourself, but it will remind you of how much you have to be grateful for.

***Greatest achievement/proudest moment so far..***

Getting married and having children is the best thing that has happened to me (besides receiving the sacraments and praying). I regard it as a miracle because it's something that I never thought would happen. It was like starting a new, better, and more real life.

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

I have completed a lot of higher education. I have BA in psychology, an MA in experimental psychology (where I studied BCM for 2 years), a JD, and an LLM in biotechnology and genomics.



All donations we collect go exclusively to fund the most promising medical research which have as their purpose the treatment or the cure of BCM.

**Donate**



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