

Spring Newsletter 2024

BCM Families Foundation <info@bcmfamilies.org>

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



Dear Friends and Families,
Happy Spring and Happy Easter!
I'm reporting advancements in the BCMFF's projects:

The clinical project of Drs. Artur Cideciyan and Tomas Aleman, of the University of Pennsylvania, is going to deliver us a clinical protocol and

outcome measures for the clinical trial. We are very excited and we hope to receive all documents soon.

University of Pennsylvania has asked us an extension of the project, to involve a European clinical center in the test of the outcome measures. We will need to deliver \$35,000 more during 2024.

At the West Virginia University, Dr. Wen Tao Deng is still working on testing AAV vectors for gene therapy, using animal models developed by BCMFF in collaboration with our Scientific Advisory Board.

The project to automate DNA tests began, in collaboration with the Institute of Molecular and Clinical Ophthalmology Basel (IoB) - Basel, Switzerland alongside the University of Tübingen is going ahead and we are waiting to receive results.

An important news is coming from Blue Gen Therapeutic Foundation: at the end of February, John Cavitt released the Press Release about Gene Therapy for BCM that you can [read here](#) , telling us that he secures exclusive license for promising Gene Therapy programs from Adverum Biotechnologies:

"We are grateful to BCMFF and Adverum for their support and guidance as we seek to move this program so many tirelessly invested in to clinic trials. Though the future can never be certain, our unified goal of achieving a safe and efficacious treatment for BCM made a big step in the right direction. This brings joy and optimism to us as parents; we hope it brings the same to your family as well." -John and Leah Cavitt

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1) BCM Families Foundation's Tenth Anniversary: Celebrating a Decade of Progress

The BCM Families Foundation celebrates its tenth anniversary.

Over the past 10 years, we've fostered a community of support and ignited

collaboration among top-tier researchers, restarting scientific research on Blue Cone Monochromacy.

As we celebrate this milestone, we are filled with gratitude for the ongoing efforts of our supporters and we are determined in our commitment to move forward.

Our story begins in 2014, and it is the story of a community of patients, who are fighting for Blue Cone Monochromatism to become a treatable disease. But the story begins long before 2014, within each of the families who are part of our community. It is the story of our maternal grandparents suffering from this form of low vision whose name they did not know before 1980s or of others before them, or of our father, of a brother, of a cousin or of an uncle and today of ourselves, our children and our grandchildren...

Read more about our story

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

2) Join the Patient's Registry

Blue Cone Monochromacy affects 1 person out of 100,000 and, at present, has no cure. The purpose of the BCM Registry is to longitudinally collect data from patients with Blue Cone Monochromacy internationally.

Through the BCM Registry, we will help to gain a better description of the clinical features, variations in phenotype and the natural history of this disease, meaning its characteristics, management, and long-term outcomes with or without treatment. This will ultimately support research and care.

Adults and children with genetic confirmation of BCM may participate in the

BCM Registry. The registration procedure is easy and fast. Find out more at: <https://www.bcmregistry.org/>

A comprehensive tutorial, which provides step-by-step guidance on how to enroll, is also available at the following link: https://www.youtube.com/watch?v=x4u4iKcu_Mo

3) Audiobooks

by Paul Rowland, Ph. D.

Audiobooks provide a gateway to literature for people with low vision. Skilled readers make stories come alive. Modern digital formats allow easy access to audiobooks on smartphones, tablets, and computers, in addition to the players available from the Library of Congress. Thousands of titles across many genres are available for quick download. From classics to contemporary bestsellers, audiobooks make the magic of reading available to people who have trouble reading conventional print.

Here are several ways to enjoy audiobooks:

1. **The Library of Congress BARD** system allows easy book downloading from a wide selection. This is a free service. Check with your state's library for the blind. You can also subscribe to commercial audiobook platforms like Audible, Audiobooks.com, or Libro.fm. These services offer extensive libraries of audiobooks for purchase or through subscription plans.
2. **Public Libraries:** Many public libraries offer audiobooks for borrowing. Check with your local library to see if they provide audiobooks through apps like OverDrive, Libby, or Hoopla.
3. **Online Retailers:** Retailers like Amazon also offer audiobooks for purchase.
4. **Streaming Services:** Some streaming platforms, like Spotify or Apple Music, offer a selection of audiobooks as part of their catalog.
5. **Podcasts:** While not traditional books, podcasts often feature serialized storytelling or non-fiction narratives, providing an alternative to audiobooks.
6. **Newspapers and magazines:** Some newspapers, such as the *Washington Post*, offer audio versions of most of their stories.

I download audiobooks from the Library of Congress BARD system and buy Audible audiobooks from Amazon. Our public library offers audio books online, but the loan period is often too short.

Audiobook in cars often involve families with children. When my children were young, our family enjoyed the Lemony Snicket series and the Harry Potter

books. Anthony Horowitz is a writer of interesting books for adults, but also has some juvenile fiction, such as the Alex Rider series.

My wife and I have enjoyed detective series by Charles Todd, Louise Penny, and William Kent Krueger.

Longer car trips can be boring, since I'm unable to see much of the detail along highways, especially Interstates. Listening to Talking Books makes the ride much more interesting.

4) **60 seconds with... An interview with Bradley Harrison**

by Trudi Dawson



Name: Bradley Harrison

Age: 33

Where do you live?

Edmonton Alberta, Canada

How many relatives do you have with BCM?

Possibly one, my nephew who is 5.

What is your job?

I am a warehouse manager for Gordon Food Service.

What are your hobbies?

I love to travel, I have been to 17 countries, I like to snowboard in the winter, watch movies and play video games.

What is your most useful BCM tip?

Utilise apps on your browser to change the colours or contrast to see better.

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

Don't be afraid to ask for help in school or seem like the odd one out. I avoided that when I was a kid because I didn't want to be seen as different, but it's something I regretted as I got older. Just because you have trouble seeing, doesn't mean you can't get a good education and pursue your dream career.

Greatest achievement/proudest moment so far...

I would say my greatest achievement is getting to where I am today in my career, and not letting my vision problems stop me from being successful.

It's easy to sit and feel sorry for yourself, and think oh I can't drive so I can't do anything. But there is always a way, and although I didn't do well in school I was able to turn it around and get a good career.

Not many people know this about me but..

I'm obsessed with learning about the universe and our solar system and have the planets tattooed on my arm.



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

Donate

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