

Holiday Newsletter 2023

BCM Families Foundation <info@bcmfamilies.org>

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Dear Families,

2023 has been a year full of projects and emotions. At the end of April in Charlotte, NC, we had a wonderful families' meeting and in mid-September in Tübingen, Germany, we had an European meeting together with all achromat associations. You will find

photos and stories of the Tübingen meeting below in this Newsletter. This year we initiated several important projects. At the University of Pennsylvania, a clinical project started with Drs. Artur Cideciyan and Tomas Aleman, who took over the Center for Hereditary Retinal Degenerations after the passing of Dr. Samuel Jacobson. We have more to say below in this Newsletter. The aim of the project is to define endpoints and to write a protocol for the clinical trial. At the 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. Finally, a 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.

Below, you can read about new arrivals among the directors, ambassadors and scientists who are part of our community. We are very happy and satisfied with all our projects, both those to meet each other and build our community of people, and the scientific ones to arrive at a cure. I am very happy to have seen a community of active and aware people this year, able to promote and support so many projects!

Renata Sarno, Ph.D.
President
BCM Families Foundation



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1) News from the Board of Directors – Welcome to our new director, Ned Reade

Since October 2023, Ned Reade is a Director of the BCM Families Foundation. His family counts multiple members affected by BCM and he is deeply interested in looking for a cure for BCM. Read more here <https://www.blueconemonochromacy.org/bcm-families/>



2) News from the Board of Ambassadors – Welcome to our new Germany Ambassador, Julius Bolay

Julius lives in Stuttgart, Germany, is a railroad engineer (Dipl.-Ing.) at Deutsche Bahn and is 27 years old. Julius' vision is to raise social awareness of the disease, to encourage affected people to pursue their professional and private goals despite illness and disability and to promote a cure for BCM. Find more here <https://www.blueconemonochromacy.org/ambassadors/>



3) News from the Scientific Advisory Board – Artur Cideciyan is a new member

Dr. Artur Cideciyan has become a member of the BCMFF's Scientific Advisory Board as of July 2023. He is a Research Professor of Ophthalmology and co-Director of the Center for Hereditary Retinal Degenerations at the Scheie Eye Institute of University of Pennsylvania.



4) A special thanks to everyone who donated!

We would like to express our appreciation for every person who took part in the fundraising efforts on Giving Tuesday 2023.

The fact that Blue Cone Monochromacy is so rare, estimated to affect 1 in 100,000 people, has been extremely challenging when it comes to raising funds.

This is why each one of your generous donations is instrumental for our mission to achieve a #Cure4BCM.

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5) A brief account of the Tübingen Meeting

We had really a good time in Tübingen, a small city full of attractions.

We met Jo Honeyman with her son, other German families with BCM, among them Julius Bolay, who was interviewed during the meeting.

Among the speakers, Prof. Jean Bennett from the University of Pennsylvania, a pioneer of gene therapy, gave an impressive lecture. Dr. Susanne Kohl made an outstanding presentation too. There were other patient organizations coming from several European countries and a lot of children.

We Blue Cone Monochromats have donated samples of blood for a new project of computerized DNA tests (NGS WES).

In the evening there were outside tables in the city centers and Oktoberfest events.



Dr. Susanne Kohl's presentation



Our president, Renata Sarno, with the Italian Achromatopsia Association president
Elisabetta Leonardi Luchetta



Dr. Bernd Wissinger and Dr. Susanne Kohl with patients testing their DNA or donating blood samples for research

6) Update on the Center for Hereditary Retinal Degenerations (CHRD) Scheie Eye Institute, University of Pennsylvania

We decided to write about the [CHRD](#) and its Founding Director, Dr. Samuel G. Jacobson, who had a close relationship with our President, Dr. Renata Sarno, for more than ten years. Before Dr. Jacobson's untimely passing in January 2023, he had been working on Blue Cone Monochromacy, first with the support of our President, and later with the support of the BCMFF. CHRD is now co-directed by Drs. Artur V. Cideciyan, PhD and Tomas S. Aleman, MD. The [Center](#) is a clinical facility that provides diagnostic evaluation and clinical consultation for patients with inherited retinal diseases (IRDs) such as Blue Cone Monochromacy, achromatopsia and many others, and performs research to better understand the basis and progression of these conditions.

Co-director Dr. Aleman provides clinical care to IRD patients who carry the burden of progressive vision loss that is currently not treatable. Co-directors Dr. Cideciyan and Dr. Aleman perform

world-class research in clinical trials and in the laboratory to make progress towards treatments. At CHRD there are several clinical trials that are either currently ongoing or soon to be starting to enroll patients.

Dr. Tomas S. Aleman, MD, is a board-certified ophthalmologist and a fellowship-trained medical retina and retinal degeneration specialist with a distinguished 30-year career in inherited retinal diseases, and provides exceptional clinical care for individuals with IRDs. Backed by a dedicated team and state-of-the-art equipment, Dr. Aleman offers a comprehensive range of services including clinical and genetic diagnoses, second opinions, routine, and specialized care. His expertise and commitment to patient care have earned him recognition as a leading authority in the field.



Dr. Tomas S. Aleman, M.D.



Dr. Samuel G. Jacobson and Dr. Artur V. Cideciyan

Samuel G. Jacobson Memorial Research Fund was established

to support some of the cutting edge work the Center does towards eradicating inherited retinal diseases.

7) Holiday gifts by Dean Monthei



With the holidays approaching it was suggested I write about holiday gifts for kids with BCM. Magnifiers and close focus monoculars are what every BCM kid should have, holiday or not.

Good pocket magnifiers available on [opticsplanet.com](https://www.opticsplanet.com) include:

Bausch & Lomb Packette Magnifier Loupe 81-31-33 - \$10

Bausch & Lomb Folding Pocket Three-Lens Magnifier Loupe 81-23-67 - \$30

Good low-cost close focus monocular available on Amazon:

Carson CloseUp 6x18mm Close-Focus Monocular (CF-618) - \$19

I recommend everyone with BCM or parents get printed catalogs from LS&S Products and Maxiaids ([lssproducts.com](https://www.lssproducts.com) and [maxiaids.com](https://www.maxiaids.com)). Printed catalog request links are at the bottom of both home pages. They both have good websites but a catalog is much easier to browse when you do not know what you are looking for. It would also be good to let kids themselves browse through the catalogs to know what is

available.

Another thing to do is search on Amazon for “low vision” or search on Google for “low vision products”.

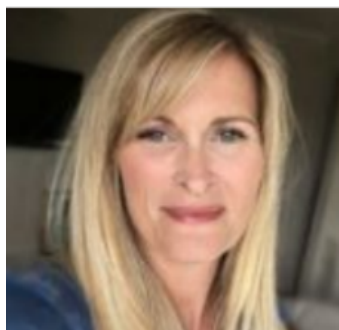
Nice-to-have gift items include:

- 1) Large-print low vision keyboards for PC or MAC (large white letters on black background)
- 2) Low vision playing cards
- 3) Large print Sudoku and crossword puzzles
- 4) Eyoyo Portable Digital Video Magnifier on Amazon - ~\$70
- 5) High contrast white on black analog wristwatch (or talking wristwatch) - ~\$25
- 6) Apple Watch - ~ \$250 to \$800
- 7) Scale with wall mount/handheld display – search Amazon for “shipping scale” - ~\$50
- 8) Eschenbach Vision Close Focus Binoculars 6X on Amazon = ~\$226
- 9) Electronic mouse magnifier (“video in” plug on TV or monitor) on Amazon - ~\$70
- 10) Acrylic Book Stand for Reading, Amasrich Adjustable Holder with 360° Rotating Base on Amazon - ~\$37

The last item on the list allows a book to be held about 10 inches above a table so it might help people neck or back problems.



8) 60 seconds with Oscar Wohlfahrt... An interview by Trudi Dawson



Name: Oscar Wohlfahrt

Age: 22

Where do you live?

I currently live and study in Copenhagen, Denmark, but I'm originally from Oslo, Norway, where I spent most of my life.

How many relatives do you have with BCM?

My younger brother Edvard also has BCM, and our grandfather had it too.

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

I am currently studying for a bachelor's degree in General Engineering. I am quite unsure of what I want to work with in the future and could see myself trying out several different jobs. I also

think it would be pretty cool to try to create my own company at some point.

What are your hobbies?

Outside of studying, rowing has been a significant part of my life. I used to be a member of the Norwegian national rowing team, competing both nationally and internationally. Currently, I'm part of a student rowing team in Copenhagen, and we're gearing up for a race in Zurich in December! Apart from rowing, I enjoy spending time with my friends and girlfriend.

What is your most useful BCM tip?

Use your phone to zoom in and take pictures when you can't see stuff. I have taken pictures of basically everything that has been written on a blackboard during my university lectures. It's helpful for seeing what the lecturers have written, but also works well as notes!

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

I would tell younger BCM boys, including my younger self, not to fear challenging situations related to their eyesight. With time, you'll find clever ways to overcome these challenges. While it might be uncomfortable initially, it gets easier, and eventually, you won't think about BCM-related issues.

Greatest achievement/proudest moment so far:

One of my proudest moments was deciding to study abroad. Despite the potential challenges, it has been an incredible experience. I've made new friends, learned a lot, and broadened my horizons.

Not many people know this about me but...

Most people I know aren't aware that I have BCM. They simply assume my eyesight is weak, but they don't know much beyond that.



Oscar Wohlfahrt

We are looking for volunteers to join our '60 Seconds Community'.
If you would like to feature here please contact
trudidawson@yahoo.co.uk, a list of questions will be sent to you
and we'll do the rest.

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