

Summer Newsletter 2023

BCM Families Foundation

Fri 8/11/2023 4:35 PM

To:renata.sarno@bcmfamilies.org <renata.sarno@bcmfamilies.org>



Dear Families, Dear Friends of the BCMFF,

I am happy again this year to reach you with the Summer Newsletter to bring you all the news and information on the activities of the BCMFF.

The Charlotte meeting, which was held on April 29, 2023, was an opportunity to meet many families and to share the path towards treatment together. We got to know Dr.

Artur Cideciyan and Dr. Tomas Aleman of the University of Pennsylvania, who are taking over all the work done by Sam Jacobson to continue it. I wanted to let you know that BCMFF has signed a research contract with the University of Pennsylvania to continue clinical studies and obtain the documents and reports useful in the event of a clinical trial for gene therapy. The funding we have agreed to give is **\$148,759**, and the timeline will end in 9 months starting from June 1, 2023. For this project we have received a donation from Mark and Diane Young for \$20,000 but we ask you to contribute with donations or with dedicated donations in order to send the funds on time.

The second project that we have agreed to finance is a project for testing some Adeno Associated viruses (AAV vectors) developed by the University of California Berkeley (UCB) and which could be injected intravitreally, i.e. without performing a surgical operation but with an injection, to carry the red opsin protein to the retina of people affected by BCM. Dr. John Flannery of UCB sent us the vectors, and Dr. Wen Tao Deng of West Virginia University (WVU), where colonies of BCM mice are maintained, has started all tests. The project for **\$130,888** plus costs for legal fees and to reserve the Intellectual Property (IP) rights to us, was fully funded by your President, Dr. Renata Sarno, for a total of \$135,000. To fully buy IP rights we will need a total of \$150,000, on November 2023.

Between now and the end of the year these will be the projects we will finance and we ask you to help us with your donations plus member volunteers searches to find grant funding.

We have many other projects related to diagnosis and clinical studies, for example we would like to carry out a Natural History clinical Study in Europe, we would like to finance a project to automate DNA tests for BCM, we would like to continue the Videogame project for diagnosing BCM and Achromatopsia, we would like to create an online CME certified course for general practitioners and pediatricians for the knowledge of genetic diseases of the retina.

Anyone who wants to devote their time to these projects is welcome: we need you!

There have been some changes, as you will read below, in our Boards of Directors and Ambassadors, if you are interested in joining these boards and to donate your time and your skills, then this is a call for you, please send me a motivation letter with a short bio to renata.sarno@BCMfamilies.org

Now is a great time to solidify your commitment to the BCMFF!

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) Tübingen September 16, 2023 - European Meeting of families with Achromatopsia and Blue Cone Monochromacy

To all European families:

The third European Meeting of families with Achromatopsia and Blue Cone Monochromacy has been organized by the Germany Achromatopsia Association, and it will take place in Tübingen, Germany, on September 16, 2023.

For more information please check the following invite:



Registrierung*

The patient day will take place/ takes place in a hybrid format. Please register for participation (in presence or online/ virtual) until Friday, 8th of September 2023 at:
angelika.braun@med.uni-tuebingen.de



The organizers



**Universitätsklinikum
Tübingen**

Achromatopsie
Selbsthilfe e.V.



BCM FAMILIES FOUNDATION
www.blueconemonochromacy.org

Universitäts Augenklinik



Approach | Parking

- In Tübingen, orientate yourself by the signs ‚Kliniken Berg‘.
- Use bus lines X15, 5, 13, 17, 18, 19 and 24. The bus stop ‚Kliniken Berg‘ is about 10 minutes' walk from the eye clinic.
- The car park ‚P6‘ near the clinic is located at the end of Elfriede-Aulhorn-Straße.

How to find the lecture room:

- From the main entrance, turn left (direction to the ENT clinic) and cross the waiting area.
- Turn right towards the stairs. Go downstairs one floor.
- Follow the signs ‚Lehrzentrum‘. They will lead you to the lecture hall.

III. European Patient Meeting on Achromatopsia and Blue Cone Monochromacy

Saturday, 16th of September 2023 | Hybrid
 Universitäts-Augenklinik Tübingen | Auditorium
 Elfriede-Aulhorn-Str. 7, 72076 Tübingen

* By registering/ with your registration, you agree that your data will be used within the hospital for organizational purposes and for the creation of the guest list. The data protection guidelines of the UKT apply to the storage of the data within the hospital (www.medizin.uni-tuebingen.de). You have the possibility to object the storage and transfer of your data at any time. The organizer must be informed/ advised of the objection in written form (see contact details, Registration).

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Responsible for content
 Dr. med. Tobias Peters

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**Universitätsklinikum
Tübingen**

III. European Patient Meeting on Achromatopsia and Blue Cone Monochromacy

We warmly welcome you to this special patient day in Tübingen! In the context of the **BMBF project Colour Bridge*** the **Achromatopsie Selbsthilfe e.V.**, the **BCM Family Foundation** and the **University Hospital Tübingen** have joined forces as organizers. International guests give lectures on medical, scientific and practical topics. Patients report on how they cope with life and answer questions. The Tübingen project leaders of the **Colour Bridge* gene therapy study** will present the results of the study for the first time.

Between the lectures there will be time for a lively exchange of ideas. On-site childcare is available for your children and we also provide a delicious catering during the event.



Hans-Werner Merkelbach
Achromatopsie Selbsthilfe e.V.



Renata Serano
BCM Family Foundation



Dr. med. Tobias Peters
Dep. für Augenheilkunde




Bundesministerium
für Bildung
und Forschung



Programme

SATURDAY, 16th OF SEPTEMBER 2023

<p>09:00 Welcome</p> <p>09:15 Presentation of the European patient organizations</p> <p>09:45 Achromatopsia and Blue Cone Monochromatism: Genetic causes and functional basis Dr. Susanne Kohl Tübingen, DE</p> <p>10:30 BREAK</p> <p>11:00 Achromatopsia and Blue Cone Monochromacy - from Laboratory Research to Gene Therapy Prof. Dr. med. Jean Bennett Philadelphia, USA</p> <p>12:00 LUNCH BREAK</p> <p>13:30 Panel discussion with patients: "My Life with Achromatopsia and Blue Cone Monochromacy - "Difficulties, Opportunities and Hope". Moderation: Dr. med. Tobias Peters Tübingen, DE</p>	<p>14:30 Update on Colour Bridge Study and first clinical Results (Part 1) Dr. med. Tobias Peters Tübingen, DE</p> <p>15:00 BREAK</p> <p>15:15 Colour Bridge-Study - First Clinical Results (Part 2) Prof. Dr. med. Katarina Stingl Tübingen, DE</p> <p>15:45 colorADD - the color alphabet Miguel Neiva Porto, Portugal</p>	<p>■ Accompanying programme: Information on aids; Possibility of genetic testing, a.o.m.</p> <p>■ Friday, 15.09.2023, 7 pm, Youth Hostel Tübingen: Evening of encounter with guests of European achromatopsia and BCM groups</p>
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2) News regarding the Board of Directors

On April 5, 2023 John Cavitt resigned from the Board of Directors.

We are all grateful to him for more than 5 years of intense activity and his commitment to support the mission of the BCM Families Foundation.

He will now move on to pursue other missions. We wish him well on his future endeavours.

Board of Directors

3) News regarding the Board of Ambassadors

As of June 2023, Trudi Dawson became President of the Board of Ambassadors.

We wish her to be successful in leading this important mission.

Here follows a message from Trudi Dawson herself:

"I am so honoured to be asked to lead the Ambassadors team for BCM Families Foundation. I found BCMFF shortly after my son was born and diagnosed with BCM and I'm not sure what I would have done without the information provided at the time. But of particular benefit to my family has been the community and connection, friendships and support I have found here. I never pretend to understand at a deeper level the science behind the cure we are working towards, but I do have a passion for supporting each other and building a resilient and connected community. If you are interested in becoming an Ambassador for BCM, or would like to just say hello and get to know me a little more, please feel free to drop me a line. I'd be happy to connect with more of our BCM Family at any time. "

To contact Trudi Dawson please write an email to: info@BCMFamilies.org



Trudi Dawson, President of the BCMFF Ambassadors' Board.

Board of Ambassadors

4) Travel tips for BCM solo travelers

by Dean Monthei



I traveled solo a lot for work to about 12 different countries. Some travel tips are discussed below along with a tip for iPhone users.

Close focus monoculars or binoculars are useful for students but are also key for traveling solo. In airports, the video displays showing flight arrivals and departures are often up high. Today you can often take a cell phone photo and magnify the photo to read these displays. Before cell phones, I used a monocular to read arrival/departure signs and the signs at the gate. Monoculars can also be used for restaurants with menu boards on the wall behind the counters. Monoculars are also useful to read the seatbelt signs, button labels and the infotainment system.

Today you can also check flight info on your phone using the airline or airport apps. There are also airport maps readily available online that show restroom, restaurants, taxi locations, etc.

Colors can also be a problem when traveling . Some airplane seat map apps or websites use color combinations on maps that are unreadable for BCMers. In some cases, a cell phone can help by using the grayscale filter accessibility function for live images or photos. This is especially helpful for red on black signage (elevators with red LEDs, etc.). The red gets converted to medium gray to distinguish it from black. In cases where two colors are hard to distinguish, converting to grayscale will sometimes help. If that all fails, ask someone for help.

For iPhone users, there is also now a free app called OKO that reads crosswalk signs and vibrates the phone in specific ways to indicate when you can walk. Here are 2 videos on OKO:

OKO Overview

Usage details

For moms vacationing with your BCM children this summer you can use this as an opportunity to get your kids to be more independent and confident by asking them to help read signs, etc. using their monocular to help out and/or putting airline apps on their phones for them to check.

Recommendation for short focus monocular (~\$20):

Amazon Link

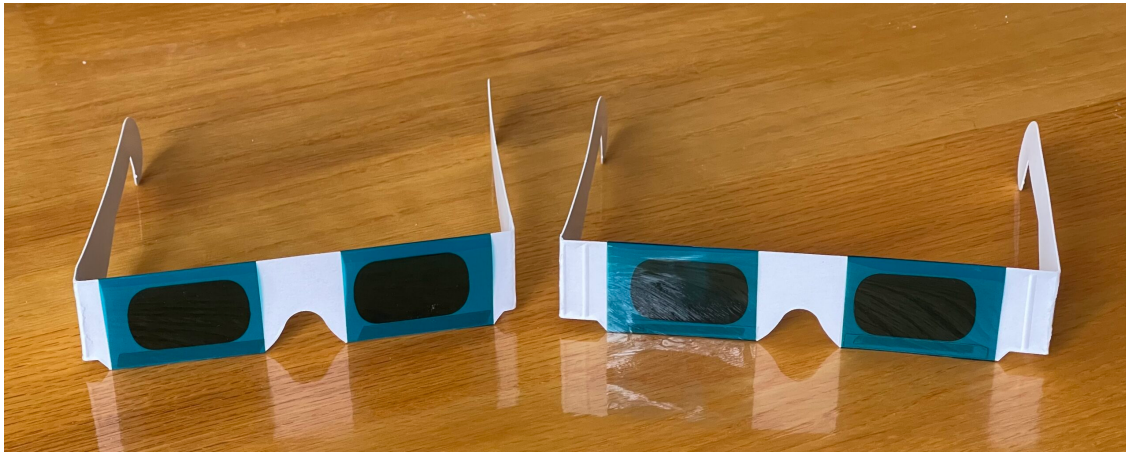


5) **BCM simulation glasses** by Dean Monthei

BCM simulation glasses are available for normally sighted family members, teachers and doctors to get a better understanding of what BCM color vision and worst case BCM acuity is like.

Click the button for a detailed description of the glasses and how to request them free of charge:

BCM simulation glasses



6) 60 seconds with... An interview with Nacer Boussahoul

by Trudi Dawson



60 seconds with.....

Name: Nacer

Age: 31

Where do you live?

Bordeaux, France.

How many relatives do you have with BCM?

My 3 brothers.

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

I am an Electronics Engineer.

What are your hobbies?

I love hiking, being lost in wild spaces. I really enjoy martial arts practice and recently discovered a passion for playing piano.

What is your most useful BCM tip?

One obvious one, but so essential - use your phone camera to replace the things your eyes cannot see; Unable to see which sandwiches are available in the storefront of the bakery? Take a picture and zoom in! Can't recognize the color of

a garment? There is an app to tell you. Small characters on a paper letter? Use your phone as a magnifier! Today, we have the great chance to all have a powerful technological instrument in our pocket that can be so helpful.

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

If only I could have realised this earlier, I would love that every young BCM affected person stops being afraid, ashamed, and angry because they are different. Sometime, people will mock you, sometimes every day little things will be a harder with your eyes than for the others, but these are just challenges which will make you stronger. Do not see BCM as a weakness, but instead as a strength, doing harder things everyday will allow you to do amazing things you thought impossible later. You have a superpower inside you, but you just don't see it yet.

Greatest achievement/proudest moment so far..

Just proud of being where I am today when looking where I come from. Proud of having finished my studies and doing a job I am passionate about, despite the fact I have been told when I was younger that it would be impossible with BCM.

Not many people know this about me but..

I still don't know how to swim! And at 11 years old I ran away from home and police had to look for me because I was waiting Hagrid to take me to Hogwarts.



All donations that 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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