BCM
FAMILIES
FOUNDATION
Toward the cure of Blue Cone Monochromacy
PO Box 7711 - Jupiter FL 33458-7711 USA

BCM Families Foundation

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BCM Families Foundation, BCMFF, is a non-profit **501-(c)-3** organization dedicated to a rare genetic disease of the eye's retina, Blue Cone Monochromacy (BCM), similar to Achromatopsia.

The **Mission** of BCMFF is to eradicate BCM by supporting the most promising biomedical research that will ultimately lead to a cure.

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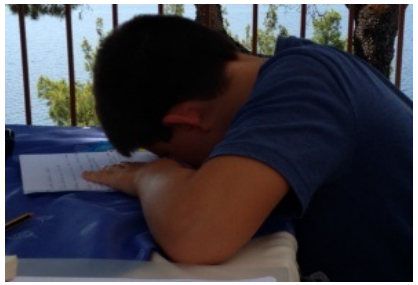
Blue Cone Monochromacy is a genetic X-linked retinal disorder that affects 1 in 100,000 individuals. Only males are affected.

Symptoms:

- poor central vision with a visual acuity of 20/400 to 20/60,
- intolerance to light (hemeralopia, photophobia),
- poor or no color discrimination,
- infantile nystagmus or nystagmus (involuntay rhythmic, oscillating motions of the eyes),
- myopia or high myopia,
- nearly normal retinal apparence,
- stable condition present from the birth,
- Systemic Features: none.



A child with BCM cannot read easily

Other Names: Blue Cone Monochromatism, BCM, X-Linked Achromatopsia
Information: OMIM 303700
Genetics: Inheritance: X-linked Recessive; Location: Xq28; Genes: LCR, OPS1LW, OPS1MW.

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BCMFF Collaborations

University of Pennsylvania – Dr. Samuel Jacobson, Dr. Arthur Cideciyan
University of Florida – Dr. William Hauswirth, Dr. Jijing Pang
University of Tuebingen, Germany – Dr. Bernd Wissinger, Dr. Susanne Kohl, Elena Buena-Atienza
AGTC, Applied Genetic Technology Corporation

BCMFF Financed Projects toward gene therapy (more than $ 2 mil)

- 2010-2014 Private funds: $1,250,000 to start gene therapy projects at University of Pennsylvania and University of Florida
- 2014-2017 BCMFF grants: $700,000 to University of Pennsylvania and University of Florida
- 2010-2016 Private funds + BCMFF grants: $220,00 to University of Tubingen

Main goal is to submit an IND* to FDA and EMA for a Phase I/Phase II gene therapy Human Clinical Trial.

*IND= Investigation New Drug

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University of Pennsylvania – Dr. Samuel G. Jacobson, Dr. Arthur V. Cideciyan

Since 2010 a collaboration started with University of Pennsylvania. The aim of this collaboration is to understand if gene therapy can be a treatment for Blue Cone Monochromacy. Positive results, never obtained before, show the presence of cone cells in the BCM patients. These cells can be treated with gene therapy. Inclusion/exclusion criteria and outcome measures are currently under investigatios.

Publications obtained with the BCMFF's support:

HUMAN GENE THERAPY 24:993–1006 (2013) 'Human Cone Visual Pigment Deletions Spare Sufficient Photoreceptors to Warrant Gene Therapy'.

PLOS ONE | DOI:10.1371/journal.pone.0125700 (April 2015) 10(4):e0125700 'Blue Cone Monochromacy: Visual Function and Efficacy Outcome Measures for Clinical Trials'.

IOVS (June 2016) 57(7):3211-21 'Developing an Outcome Measure With High Luminance for Optogenetics Treatment of Severe Retinal Degenerations and for Gene Therapy of Cone Diseases '

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University of Florida – Dr. William W. Hauswirth Dr. Jijing Pang

Since 2010 a collaboration started with University of Florida. Initially (2010-2014) thanks to private funds and then as a part of a Sponsored Research Agreement with BCMFF, Dr. William Hauswirth developed, for the first time, an animal model of BCM and treated it with an AAV-based gene therapy. Superb positive results show the possibility to consider gene therapy as a treatment of BCM. Scientists are currently involved in testing different kind of vectors.

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University of Tuebingen, Germany – Dr. Bernd Wissinger, Dr. Susanne Kohl

Since 2010 a collaboration started with the aim to identify all possible BCM causative mutations. Patients without a genetic confirmation of their disease have been invited to test their DNA.

Main objectives of the project are:

- Development of sensitive and reliable methods for the detection of mutations and structural aberrations in the opsin gene cluster
- Development and maintenance of Standardized Operation Procedures for the genetic testing for BCM
- Reference center for the genetic testing of clinically diagnosed BCM patients
- Provision of reference material for quality control in genetic testing for BCM
- Determination of mutation origin and new mutation rates in the opsin gene cluster

http://www.eye-tuebingen.de/wissingerlab/projects/blue-cone-monochromacy/

Publications:

Elena Buena-Atienza et al. 'De novo intrachromosomal gene conversion from OPN1MW to OPN1LW in the male germline results in Blue Cone Monochromacy' www.nature.com SCIENTIFIC REPORTS 6:28253 DOI: 10.1038/srep28253

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AGTC, Applied Genetic Technology Corporation

Applied Genetic Technologies Corporation (Nasdaq:AGTC), a biotechnology company conducting human clinical trials of adeno-associated virus (AAV)-based gene therapies for the treatment of rare eye diseases, and the BCMFF started in 2016 a collaboration to develop an AAV-based gene therapy for BCM.
AGTC has deep expertise in the optimization and manufacturing of novel AAV-based therapies for the treatment of inherited retinal diseases. As part of the collaboration, the BCM Families Foundation will share with AGTC scientific and clinical study data that have been generated through its research efforts, and will also provide patient advocacy and clinical trial support to AGTC. AGTC will be responsible for product development efforts and aspects of the clinical program design and execution.
AGTC's lead product candidates focus on inherited orphan diseases of the eye, caused by mutations in single genes that significantly affect visual function and currently lack effective medical treatments. AGTC's product pipeline includes six named ophthalmology development programs across five targets [x-linked retinoschisis (XLRS), x-linked retinitis pigmentosa (XLRP), achromatopsia, wet age-related macular degeneration and now blue cone monochromacy], one non-ophthalmology program (alpha-1 antitrypsin deficiency) and proof-of-concept data in multiple additional indications.

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BCMFF Board of Directors

Dr. Renata Sarno, President

Renata Sarno, Ph.D., has 5 relatives with BCM, including her sons. In 1992 she earned a Ph.D. in Theoretical Physics, then worked in research about supercomputing and sub-nuclear physics until 1994. From 1994 to 2008 she & 3 other shareholders founded a web company, venere.com, the first on-line hotels reservation system gathering more than 20,000 hotels especially in Europe and employing more than 200 people. She sold Venere to Expedia in 2008. Dr. Sarno created a BCM Families website that has developed international contact among families coping with Blue Cone Monochromacy (BCM)., Dr. Sarno is a committed advocate for BCM cure. She is on the Board of Directors of Associazione Acromati Italiani, a non-profit organization for Achromatopsia and BCM in Italy, where she lives.

Kay McCrary, Secretary

Kay McCrary, Ed.D., whose father, uncle, great-uncle, & 4 male paternal cousins, have BCM, is now grandmother to 3 cherished boys who are legally blind due to BCM. She is highly motivated to improve their lives. Dr. McCrary has retired after 30 years as Director of Patient & Family Education at the South Carolina Department of Mental Health. She authored four books that explain schizophrenia, schizoaffective disorder, & bipolar affective disorder in layman's language & teach recovery strategies to patients & their involved families. Her workbooks have been translated into 28 different languages.

Barbara Sergent, Treasurer

Barbara Wade Sergent holds an MBA from Cleveland State University and held various management positions in the banking industry until she put her career on hold to raise her five children. Mrs. Sergent is also the founder of WadeSergent Consulting, LLC through which she has provided project management services for some of the largest banking institutions in the United States and globally. Mrs. Sergent's father, son, nephew, cousins and several uncles have BCM. Her goals are to improve the lives of those affected with BCM, find a cure and eliminate the genetic abnormality so her four young daughters do not pass on BCM to future generations.

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BCMFF Scientific Advisory Board

BCMFF has a **Scientific Advisory Board (SAB)** composed of the primary scientists worldwide who are contributing to the research about BCM. They are leading experts in gene therapy, ophthalmology, clinical investigations and molecular genetics of inherited eye diseases:

Dr. Jeremy Nathans, discovered in 1989 genes and the main genetic mechanisms causing BCM;

Dr. William W. Hauswirth, University of Florida;

Dr. John G. Flannery, Berkeley University;

Dr. Samuel G. Jacobson, University of Pennsylvania;

Dr. Alessandro Iannaccone, University of Tennessee;

Dr. Bernd Wissinger, University of Tuebingen – Germany.

Scientists in the SAB are expert and passionate about BCM.

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Other collaborations

We are working with several achromats and patients' associations in order to promote the diagnosis of Achromatopsia and BCM. Between them:

- Achroma Corp.
- Associazione Acromati Italiani onlus

With the last one we organized an Italian Network for the diagnosis of Achromatopsia and BCM.

BCMFF is part of **TRAIN, The Research Accelerator and Innovation Network** of FasterCures.

BCMFF is listed on **GuideStar**, earning its prestigious **Silver Medallion**

BCMFF is part of **Orpha.net**

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Ongoing Projects:

BCMFF **connects more than 200 patients** affected by BCM and their families.

- Educational
- Awareness and Advocacy
- Diagnosis
- Support
- Epidemiology
- Patient Registry

A child with BCM during a clinical examination

These projects are performed as part of day by day activities by Directors and Volunteers. **A Patient Registry for BCM is one of our main goal.**

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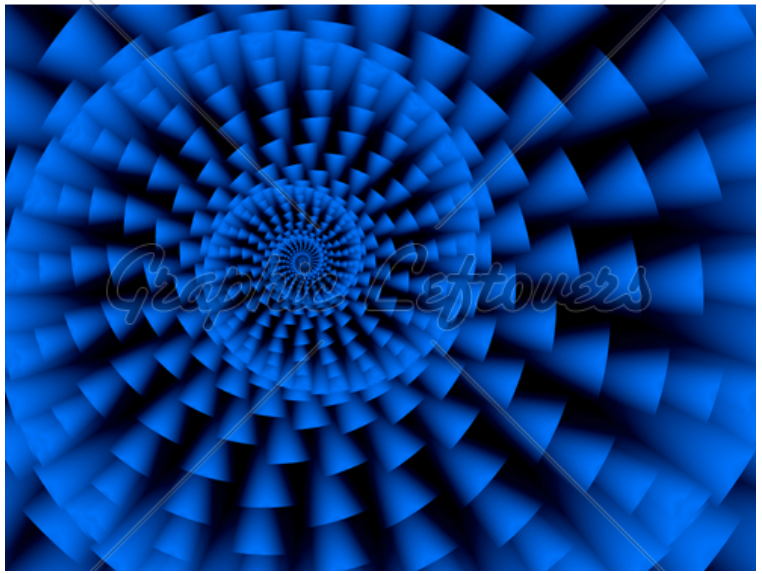
Information

Web: www.bcmfamilies.org
Forum: bcmfamilies.ning.com
Facebook: group 'BCM'
Twitter: @BCM_Families



Contact

Dr. Renata Sarno, President
Email: renata.sarno@BCMFamilies.org
76 Viale Tito Livio – 00136 – Rome – Italy
Phone: +39 335 7109560

BCM Family Foundation is recognized as a non-profit organization under 501-(c)-3 by Internal Revenue Service. Tax Identification Number 47-132773