IRB Approved: 07-Oct-2019

UNIVERSITY OF PENNSYLVANIA
RESEARCH SUBJECT
COMBINED INFORMED CONSENT/PARENTAL PERMISSION AND HIPAA AUTHORIZATION FORM

Protocol Title:	Blue Cone Monochromacy International Patient Registry
Principal Investigator:	Samuel G. Jacobson, MD, PhD Professor of Ophthalmology Director, Center for Hereditary Retinal Degenerations Scheie Eye Institute 51 N. 39th Street Philadelphia, PA 19104 Phone: 215-662-9981 Fax: 215-662-9388
Emergency Contact:	215-662-8111
Sponsor	BCM Families Foundation

Research Study Summary for Potential Subjects

For the purposes of this consent, the word 'you' may mean 'you' or 'your child' if you are the parent of a child being invited to participate in the study.

You are being invited to participate in a research study. Your participation is voluntary, and you should only participate if you completely understand what the study requires and what the risks of participation are. You should ask the study team any questions you have related to participating before agreeing to join the study. If you have any questions about your rights as a human research participant at any time before, during or after participation, please contact the Institutional Review Board (IRB) at (215) 898-2614 for assistance.

The research study is being conducted to create an online Patient Registry dedicated to a rare genetic retinal disease, Blue Cone Monochromacy (BCM). The Registry has been created by the BCM Families Foundation (BCMFF). The purpose of the Registry is to collect data from participants with BCM in order to describe the characteristics, management and outcomes of BCM. You are being asked to participate because you have BCM and you have identified Dr. Jacobson as the physician who can verify this diagnosis.

If you agree to join the study, you will be asked to complete the following research procedures: you will enroll yourself in the online registry by creating a username, entering your contact information and DNA test report and selecting Dr. Jacobson as your physician who can validate your BCM diagnosis. By signing this consent form, you

are authorizing Dr. Jacobson to release information about your diagnosis to the Registry.

Your information will be stored in the Registry indefinitely, or until you request the removal or deletion of the data from the Registry.

There is no direct benefit to participation in this study. The information obtained from the Registry may produce useful knowledge about BCM. Additionally, with your permission, researchers may contact individuals in the Registry to participate in other studies or clinical trials. The risks involved with this study are minimal. The most common risks of participation is a breach of confidentiality.

Please note that there are other factors to consider before agreeing to participate such as additional procedures, use of your personal information, costs, and other possible risks not discussed here. If you are interested in participating, a member of the study team will review the full information with you. You are free to decline or stop participation at any time during or after the initial consenting process.

Why am I being asked to volunteer?

You are being invited to participate in a research study because you have been diagnosed with Blue Cone Monochromacy (BCM), and you have selected Dr. Jacobson as the physician who can verify your diagnosis in the BCM International Patient Registry.

You do not have to participate in any research study offered by your doctor. If you choose not to participate, there will be no loss of benefits to which you are otherwise entitled. You may also decide to discuss the study with your family, friends, or family doctor. Being in a research study is different from being a patient. As an investigator, your doctor is interested both in your clinical welfare and in the conduct of this study.

If you decide to participate, you will be asked to sign this form.

This consent form is written from the point of view of a research participant. If the parent or legal guardian of a minor or a legally authorized representative will be providing consent, the words "you" and "your" should be read as ("your child" or "the research participant").

What is the purpose of this research study?

The BCM Registry has been created by the BCMFF to collect clinical and genetic information from patients with BCM, in order to understand more about the characteristics, management, and long-term outcomes of the condition. Patient registries can be important for accelerating research into rare diseases and the development of new therapies. The information in the Registry may be used for statistical purposes, to allow researchers to anonymously contact individuals to participate in studies or clinical trials, to advance the scientific understanding of BCM, and to link data of individuals of BCM with their family members where applicable.

How long will I be in the study?

There is no end date planned for the Registry. Your information will be stored in the Registry indefinitely, or until you request the removal or deletion of the data from the Registry.

What am I being asked to do?

You will register yourself in the Registry (www.bcmregistry.org/patients). You will be asked to provide your name, contact information, date of birth, race/ethnicity, and family history. As a requirement for participation in the Registry, you will need to provide a BCM genetic testing report and select Dr. Jacobson as the clinician who can validate your diagnosis. Once Dr. Jacobson does so, your enrollment in the registry will be confirmed. Dr. Jacobson will also need to provide medical, ophthalmic, and genetic history to be compiled in the Registry.

What are the possible risks or discomforts?

The risks associated with this study are minimal. There is a risk of breach of confidentiality, if someone were to obtain illegal access to the Registry and release information contained in the Registry or modify the data in some way. There are safety measures in place to protect the personal data and to prevent this from happening, such as de-identification and encryption technology.

Risks of Genetic Testing

Genetic testing is not part of this study. However, the registry requires your results from genetic testing that was performed previously. Even without your name or other identifiers, your genetic information is unique to you. The researchers believe the chance that someone will identify you is very small, but the risk may change in the future as people come up with new ways of tracing information.

Very rarely health or genetic information could be misused by employers, insurance companies, and others. For example, it could make it harder for you to get or keep a job or insurance, or life insurance companies may charge a higher rate based on this information. We believe the chance these things will happen is very small, but we cannot make guarantees.

A federal law (Genetic Information Non-Discrimination Act, GINA) helps reduce the risk from health insurance or employment discrimination. The law does not include other types of misuse by life insurance or long term care insurance. If you want to learn more about GINA, you can find information about it on the internet or ask the study staff.

What if new information becomes available about the study?

During the course of this study, we may find more information that could be important to you. This includes information that, once learned, might cause you to change your mind about being in the study. We will notify you as soon as possible if such information becomes available.

What are the possible benefits of the study?
You are not expected to get any benefit from being in this research study. This study will yield knowledge about BCM and may, in some cases, lead to your involvement in other studies or clinical trials (if you provide your consent to be contacted about such studies).

What other choices do I have if I do not participate?
You may choose not to participate in this study.

Will I be paid for being in this study?
There is no compensation for participation in this study.

Will I have to pay for anything?
There is no cost associated with participation in this study. Neither you nor your insurance company will be charged as a result of your participation in this study.

Will I receive the results of research testing?
There are no tests associated with participating in the Registry. All information provided to the Registry is information which already exists in your medical record. Therefore, no results will be returned to you.

When is the Study over? Can I leave the Study before it ends?
Your information will remain in the Registry indefinitely, or until you request to have the information about you removed by following the instructions provided on the Registry. You are free to leave the study at any time. Withdrawal will not interfere with your future care.

This study may be stopped at any time by your physician, the study Sponsor, or the Food and Drug Administration (FDA) without your consent because:

- The Primary Investigator feels it is necessary for your health or safety. Such an action would not require your consent, but you will be informed if such a decision is made and the reason for this decision.
- You have not followed study instructions.
- The Sponsor, the study Principal Investigator, or the Food and Drug Administration (FDA) has decided to stop the study.

How will my personal information be protected during the study?
We will do our best to make sure that the personal information obtained during the course of this research study will be kept private. However, we cannot guarantee total privacy. Your personal information may be given out if required by law. If information from this study is published or presented at scientific meetings, your name and other personal information will not be used. The Institutional Review Board (IRB) at the University of Pennsylvania will have access to your records. If this study is being overseen by the Food and Drug Administration (FDA), they may review your research records.

All the information about you in the Registry will be maintained in a safe and secure IT platform. Any identifying information about you will only be seen by the BCM Registry Staff and Dr. Jacobson. Other individuals, such as other clinicians and researchers, will only be able to see your medical information after any information that could identify you has been removed.

You will create a unique username and password to access the Registry and it is recommended that you do not use any identifying information while creating the username. The passwords are saved in an encrypted form to prevent anyone from accessing your profile in the Registry without knowing the password.

What may happen to my information collected on this study?

Your information could be stored and shared for future research in a de-identified fashion. De-identified means that all identifiers have been removed. It would not be possible for future researchers to identify you as the BCMFF will not share any identifiable information about you with researchers. Researchers may send communications to groups of patients about other research projects or clinical trials, if you authorize this process in the Registry, but they will not know the identity of the individuals they are contacting. This can be done without again seeking your consent in the future, as permitted by law. The future use of your information only applies to the information collected on this study.

There is a risk of breach of confidentiality (unintentional release of your information). We will do our best to make sure that this doesn't happen. However, we cannot guarantee total privacy. We will protect your confidentiality during storage and sharing by encryption technology, both in the Registry and during transfer of the data to clinicians and researchers.

If you have questions about the storage of your information, or have changed your mind, you can request the removal of your data from the Registry at any time.

What information about me may be collected, used or shared with others?

The following information may be collected, used or shared with others:

- Name, surname
- Date of birth
- Place of birth (City, country)
- Address (street, zip code, city)
- Nationality
- Race and ethnicity
- Email address
- Telephone number
- Genetic testing results
- Medical, ophthalmic, and family history

Why is my information being used?

Your information is used by the research team to contact you during the study. Your information and results of tests and procedures are used to:

- do the research
- oversee the research
- to see if the research was done right
- to evaluate and manage research functions.

Who may use and share information about me?

The following individuals may use or share your information for this research study:

- The investigator for the study and the study team
- Other authorized personnel at Penn, including offices that support research operations
- Other research personnel with access to the databases for research and/or study coordination and as otherwise approved by the IRB

Who, outside of the School of Medicine, might receive my information?

- BCM Families Foundation Registry Managers, BCMFF's SAB members, and BCMFF Registry Steering Committee's members, who can monitor and check quantity and quality of the data in the BCM Registry
- Clinicians and researchers who request de-identified aggregate data for statistical analysis
- Other patients who have requested statistical aggregated data about the Registry participants

<u>Oversight organizations</u>

- The U. S. Office of Human Research Protections (OHRP)

Once your personal health information is disclosed to others outside the School of Medicine, it may no longer be covered by federal privacy protection regulations.

The Principal Investigator or study staff will inform you if there are any additions to the list above during your active participation in the trial. Any additions will be subject to University of Pennsylvania procedures developed to protect your privacy.

How long may the School of Medicine use or disclose my personal health information?

Your authorization for use of your personal health information for this specific study does not expire.

Your information may be held in a research database. However, the School of Medicine may not re-use or re-disclose information collected in this study for a purpose other than this study unless:

- You have given written authorization
- The University of Pennsylvania's Institutional Review Board grants permission

- As permitted by law

Can I change my mind about giving permission for use of my information?

Yes. You may withdraw or take away your permission to use and disclose your health information at any time. You do this by sending written notice to the investigator for the study. If you withdraw your permission, you will not be able to stay in this study.

What if I decide not to give permission to use and give out my health information?

Then you will not be able to be in this research study.

You will be given a copy of this Research Subject HIPAA Authorization describing your confidentiality and privacy rights for this study.

By signing this document, you are permitting the School of Medicine to use and disclose personal health information collected about you for research purposes as described above.

Who can I call with questions, complaints or if I'm concerned about my rights as a research subject?

If you have questions, concerns or complaints regarding your participation in this research study or if you have any questions about your rights as a research subject, you should speak with the Principal Investigator listed on page one of this form. If a member of the research team cannot be reached or you want to talk to someone other than those working on the study, you may contact the IRB at the number on page one of this form.

When you sign this form, you are agreeing to take part in this research study. This means that you have read the consent form, your questions have been answered, and you have decided to volunteer. Your signature also means that you are permitting the University of Pennsylvania to use your personal health information collected about you for research purposes within our institution. You are also allowing the University of Pennsylvania to disclose that personal health information to outside organizations or people involved with the operations of this study.

A copy of this consent form will be given to you.

_____	_____	
Name of Subject (Please Print)	Signature of Subject	Date
_____	_____	
Name of Person Obtaining Consent (Please Print)	Signature	Date

For subjects unable to give authorization, the authorization is given by the following authorized subject representative:

Authorized subject representative **[print]**	Authorized subject representative Signature	Date

Provide a brief description of above person authority to serve as the subject's authorized representative.