BCM FAMILIES FOUNDATION
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Blue Cone Monochromacy: how to test your DNA

If you have a clinical diagnosis of BCM, there are many reasons to perform a DNA test: to have a genetic confirmation of the diagnosis, to help scientific research to find all the possible genetic mutations that lead to disease, to know possible experimental therapies that often depend on the particular genetic mutation that you have.

The genetic confirmation is important because a person with an inherited eye disease needs to understand how the disease spreads within his own family and what are the chances of passing the disease to his children.

The laboratories that perform the DNA test can be clinical or research laboratories:

Clinical

A clinical laboratory examines specimens and reports results to healthcare providers for the purpose of diagnosis, prevention, or treatment. In the United States, laboratories performing clinical tests, the results of which can be used for treatment, must be CLIA (Clinical Laboratory Improvement Amendments) approved.

Research

A research laboratory examines specimens for the purpose of better understanding a medical condition and or developing a clinical test. Some research laboratories will obtain CLIA certification (in US) so their research findings can be shared with study participants.

University Hospital Tübingen – Molecular Genetics Laboratory:

Contacts:

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Instruction on how to send blood samples to University Hospital Tübingen:

1) Read Jessica Guide below;
2) Prepare consent forms and shipping documents;
3) Prepare your clinical reports and family pedigree;
4) Contact Dr. Wissinger forwarding him your clinical reports;
5) Take contacts with an express courier able to ship to Europe;
6) Select a local practitioner able to withdrawl blood samples;

7) Prepare shipping materials.

For the DNA test, Tübingen laboratory needs 2×5 ml of venous blood for each individual, drawn in EDTA containers, that should be shipped by courier.

Blood samples can be drawn by a local practitioner. For preservation of the blood samples, it is recommended the shipment under cooled conditions.

Easiest way for that is to use a regular foam box with some cool packs. Blood samples should not be frozen, therefore please wrap the blood containers in some tissue before adding them to the foam box. In order to avoid dripping in case of breakage of the tubes it is recommended to wrap the tubes in a plastic casing.

With the blood samples it is usually requested to send a signed consent form for each individual (for minors we would require the signature of both parents), copy of identity documents for each individual, Family's Pedigree, medical examination report and to fill and sign other forms needed for the export.

In some case the result arrives after 3-4 months. In other cases more time is needed.

Where to ship the samples:

Dr. Susanne Kohl
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E-mail: susanne.kohl@uni-tuebingen.de
Molekulargenetisches Labor
Universitätsklinikum Tübingen
Elfriede-Aulhorn-Str. 5-7 D-72076 Tübingen – Germany

Dr. B. Wissinger
E-mail: wissinger@uni-tuebingen.de

Before shipping samples please take contact with Dr. B. Wissinger or with the BCMFF at info@bcmfamilies.org

When declaring a blood sample for shipment, it may now also be necessary to declare a so-called HS-code, which is 30029010 for human blood samples.

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Please visit our web site for further information:

http://www.blueconemonochromacy.org/genetic-dna-test/

Jessica Guide

I'm writing this to guide anyone wanting to send blood samples to Germany for the purpose of genetic testing in regards to BCM. I had my son's blood drawn by his pediatrician and shipped to the University Hospital Tübingen. This is how I went about the process.
Renata put me in contact by email with Professor Bernd Wissinger (He will also answer any questions you have.) and she sent me 3 forms that need to be filled out to go with the blood samples. Declaration form, consent form (if a minor, parents sign), and a commercial invoice. Once I had the forms I called my pediatrician and explained that I wanted an order for my son's blood to be drawn for BCM genetic testing. I explained to them that this was very important and the testing was being done free of charge. I had to pay out of pocket for the blood draw.

The blood samples were collected in EDTA tubes. The collection is not to be centrifuged (just inversion of the tube for mixing). No freezing of the sample. Blood sample needs to be cooled until shipment. Shipment does not require cooling of the samples. 3-5 ml of EDTA blood is sufficient. I brought all this information with me and let the doctor, nurse, and phlebotomist read it. They will understand all the lingo.

After the blood was drawn I placed it in a small cooler with an ice pack. I took it straight to my local post office and explained to them what I was doing. The post office helped me immensely. I had filled out all 3 forms that Renata had sent to me the best I could before I went to the post office. The post master helped me fill out the rest of the forms that had any missing information. The samples were shipped at the post office but overnight by FedEx. There were extra forms that needed to be filled out at the post office due to blood categorized as "biohazard". The cost to ship the samples was a little more than I expected. The cost was around $70.

Once the blood samples are shipped expect to wait 3-4 months for results to come back.
I shipped my son's samples the end of July 2014 and received a letter confirming BCM the end of November 2014.

Jessica