

# PLEASE DO NOT SIGN AT HOME. MUST BE SIGNED UPON ARRIVAL

The University of Texas Southwestern Medical Center at Dallas  
Retina Foundation of the Southwest

## CONSENT TO PARTICIPATE IN RESEARCH

Title of Research: **Retinal Pathophysiology in Infants and Adults**

Funding Agency/Sponsor: National Institutes of Health -National Eye Institute Foundation  
Fighting Blindness

Study Doctors: David G. Birch, Ph.D.; Dennis R. Hoffman, Ph.D.; Karl Csaky,  
MD, Ph.D.

Research Personnel: Kirsten G. Locke, C.R.A., R.N.; Martin Klein, M.S.

You may call these study doctors or research personnel during regular office hours at (214) 363-3911.

Note: If you are a parent or guardian of a participant younger than 18 years of age and have been asked to read and sign this form, the "you" in this document refers to the participant.

### ***Instructions:***

Please read this consent form carefully and take your time making a decision about whether to participate. As the researchers discuss this consent form with you, please ask him/her to explain any words or information that you do not clearly understand. The purpose of the study, risks, inconveniences, discomforts, and other important information about the study are listed below. If you decide to participate, you will be given a copy of this form to keep.

***What is DNA?*** DNA means *deoxyribonucleic acid*. DNA is the substance in our cells which contains information we inherited from our parents and other family members. Your DNA contains "genes" which predict things like physical characteristics (eye color, hair color, height, etc.) and may also be a factor in whether you develop or are at risk of developing certain illnesses or disorders.

### ***What is genetic testing?***

Genetic tests look for naturally occurring differences in a person's genes, or the effects of specific genes. These differences could indicate an increased chance of getting a disease or condition. Genetic testing includes gene tests (DNA testing) and sometimes biochemical tests (protein testing) if it relates to a specific gene.

In gene tests, DNA in cells taken from a person's blood is examined for differences. The differences can be relatively large - a piece of a chromosome, or even an entire chromosome, missing or added.

Sometimes the change is very small - as little as one extra, missing or altered chemical within the DNA strand. Genes can be amplified (too many copies), over-expressed (too active), inactivated, or lost altogether. Sometimes pieces of chromosomes become switched, turned over or discovered in an incorrect location.

***Why is this study being done?***

This study is being done to collect information about inherited eye diseases that will be used to better understand which genes may cause diseases and help doctors in their diagnosis and treatment.

***Why am I being asked to take part in this research study?*** You are being asked to take part in this study because you (your child) have been diagnosed with an eye disease that may be inherited. Other members of your family who may or may not have an eye disease may also be invited to be part of this research, if you agree.

***How many people will take part in this study?***

About 400 people over each year will take part in this study at the Retina Foundation of the Southwest.

***What is involved in the study?***

If you agree to be in this study, you will be asked to sign this consent form and may have some or all of the following tests and procedures, which can last up to 3 hours. You may be invited to come back on another day for more tests.

***Questions:*** The investigators at the Retina Foundation of the Southwest will ask you questions about your eye condition, general health, diet and family history, especially as to the presence of eye disease. You will also be asked to provide personal information such as your date of birth, sex, and race. You may also be asked to sign "An Authorization for Release of Medical Records" form so the investigator may obtain copies of your medical records from your eye doctor.

***Visual function tests:*** To obtain information about your vision, you may be asked to perform standard clinical tests such as visual acuity, visual fields, color pictures and images for thickness measurements of the retina. Electrical signals generated within the eye (the electroretinogram or ERG) in response to a variety of light stimuli may be recorded. This test will require you to be dilated in one or both eyes. A special contact lens will be placed upon your eye to record the electrical signals. We may also take different types of images of your eyes.

***Smell Identification Test:*** You may be asked to complete a short test for measuring your sense of smell. You will release smells by scratching test-strips with a pencil and asked to choose the smell from four choices.

***Samples of Blood:*** Up to 2 ½ tablespoons (30 ml) of blood may be drawn from a vein in your arm with a small sterile needle. This is the standard method used to obtain blood for routine hospital tests.

You may be asked for a second blood sample if the research laboratory cannot process the first sample.

***Saliva sample:*** You may be asked to give a small sample of saliva. With your permission, saliva samples may be requested from other family members.

By agreeing to participate in this research, you agree to be included in this research database. Investigators may use your health information for future research on various diseases including

genetic research. However, your personally identifiable information will never be released to researchers, so they will not know who you are or be able to contact you.

***How will my samples be identified?***

Your sample will be marked with a coded identifier and will not be personally identifiable. Neither your name nor any identifying information will be given to the researchers who receive your samples. Any results collected will not be released in a personally identifiable manner, and thus no information will be given to your insurance provider, employer, family, etc. without your permission.

***How long can I expect to be in this study?***

In many genetic studies, testing of the DNA may go on for very long periods of time. This is true because we are continually finding new genes that may be involved in genetic eye diseases. Therefore, while your direct participation in this study will be over once you have completed the procedures/visit described above; the DNA isolated from your blood/cheek cell sample may continue to be studied for many years.

***Can I stop taking part in this research study?***

Yes. If you decide to participate and later change your mind, you are free to stop taking part in the research study at any time. You may ask Dr. Birch to destroy any record of your participation in this research and to destroy any sample with your name on it. You will not be asked for further information or samples. Your identity will be removed from all research records. However, the resulting data from the research will not be discarded. De-identified copies of DNA and/or growing cells made from your samples will not be destroyed.

***What will happen to the samples collected for this research?***

Your blood/saliva sample will be used to isolate DNA for genetic analysis. Part of your blood/saliva sample will be stored in a Cell Bank and will be available for research, both now and in the future. This also allows us to perform many tests without having to ask you for additional blood/saliva samples.

***How is DNA obtained?***

Cells from blood or other body materials are processed in a laboratory that has special equipment that can extract DNA and identify genes.

***How long will my samples be kept?***

We may keep your sample in the research laboratory at the Retina Foundation of the Southwest until it is all gone, becomes unusable or until they decide to discard the sample. If your sample remains stored beyond your lifetime, your sample will be used as described in this document.

***May other researchers use my sample?***

When you provide a sample for purposes of this study your sample becomes the property of the Retina Foundation of the Southwest and may be used for future studies or provided to other investigators at other medical research facilities without any identifiers.

***Who decides which research scientists may receive samples of my DNA?***

We will decide which researchers at this medical center and at other medical centers may receive samples of your DNA. Your samples may be used in other research only if the other research has been reviewed and approved by an Institutional Review Board (IRB).

***Could my sample be used for other purposes?***

No. Your samples or your DNA will only be used for research. Research tests using your sample may possibly result in inventions or procedures that have commercial value and are eligible for protection by a patent. Compensation for any future commercial developments is not available from the Retina Foundation of the Southwest, its researchers or other facilities or researchers whose research may benefit from the use of your sample. By agreeing to the use of your sample in research, you are giving your sample without expectation of acknowledgment, compensation, interest in any commercial value or patent or interest of any other type. However, you retain your legal rights during your participation in this research.

***Will the results of research tests be reported to me?***

The results of the DNA testing will not be shared with your doctor, but you can ask to be informed about the results. However, the results will not be used to plan your health care. The results of the visual function testing can be shared with you and your doctor, if you request it.

***Is counseling available if I receive the results of DNA tests?***

Yes. Dr. Birch or Kaylie Jones can tell you what test results mean.

***Is there a charge for counseling?***

No. Neither you nor your insurance will be charged for the counseling you receive at the Retina Foundation of the Southwest.

***Are there any possible benefits from receiving test results?***

If you do receive the results of tests using your DNA, you may receive information that reduces the uncertainty about the likelihood of developing a genetic eye disease and/or passing it to your children. Obtaining the results of DNA tests may help you and other members of your family plan for the future. In some cases, early treatment of a disorder that runs in the family may improve the chances of a good outcome.

***What are the risks of the study?***

**Questions:** You will be asked questions about your health, medications you take and your diet habits. However, you can skip any question that makes you uncomfortable.

**Risk of Visual Function Tests:** The contact lens placed on the eye, can, in rare situations, cause a small scratch on the surface of the eye. Excessive rubbing of your eyes after testing can also scratch your eye. You should inform the research staff if your eye is painful, especially when you blink, after testing. The scratch will heal on its own within 24 hours. To make you more comfortable, a patch may be placed over the eye to prevent you from blinking.

**Risks of Blood Drawing:** Risks associated with drawing blood from your arm include minimal discomfort and/or bruising. Infection, excess bleeding, clotting, and/or fainting are also possible, although unlikely. If you have unusual symptoms, pain, or any other problems while you are in the study, you should report them to the researcher staff right away. Telephone numbers where they can be reached are listed on the first page of this consent form.

Depending on your age, you will have a maximum of two and a half tablespoons (30 ml) of blood collected because you are in this research study.

**Collection of the Saliva Sample:** There is no discomfort or risk associated with the collection

**Stress:** You could experience stress from participating in this kind of research. Knowing that researchers have personal information about you may trouble you. If the results of DNA tests show that you or anybody else in your family may develop a genetic eye disease, you and other family members could experience serious stress after receiving such information. If you experience stress because you participate in this research, the Retina Foundation of the Southwest can help you obtain medical care to help you manage stress. You could learn that you will not have a serious medical problem, but your children (or someone else) will.

**Personal, sensitive information:** If you are not the parent of a child in your family, or if you are the parent of a child in another family, that information could be learned from DNA tests. This kind of information will not be reported to you or other family members.

**Unforeseen Risks and New Information:** There may possibly be risks to your participation in this research which Dr. Birch does not know about now. You will be told if any new information becomes available during the study that could cause you to change your mind about continuing to participate or that is important to your health or safety.

**Loss of Confidentiality:** Any time information is collected; there is a potential risk of loss of confidentiality. Every effort will be made to keep your information confidential; however, this cannot be guaranteed. For more information, please see the section called "Will my information be kept confidential?"

***Will I be contacted in the future?***

You have the option to elect to be contacted in the future in order to obtain follow-up information or to ask you to take part in more research. (A "no" answer will not disqualify you from this research )

Yes \_\_\_\_\_ initials

No \_\_\_\_\_ initials

**If you elect "yes", please keep in touch with Dr. Birch and maintain a current address and telephone number on file. Please notify Dr. Birch if your legal name changes.**

It is your responsibility to inform a child that samples of his or her DNA may be kept in a research laboratory at this medical center or possibly other medical centers. The child will not be asked to sign another consent form when he/she reaches age 18.

***What are the possible benefits of this study?***

If you agree to take part in this study, there is usually no direct benefit to you.

We hope the information learned from this study will benefit others with genetic eye diseases in the future. Information gained from this research could lead to better detection and treatment.

***What other options do I have?***

You may choose to not participate in this study. If you decide not to take part in this research study, it will have no effect on your medical care.

***Will I be paid if I take part in this research study?***

No. You will not be paid to take part in this research study. There are no funds available to pay for parking expenses, transportation to and from the research center, lost time away from work and other activities, lost wages, or child care expenses

***Will my insurance provider or I be charged for the costs of any part of this research study?***

No. Neither you, nor your insurance provider, will be charged for anything done only for this research study (i.e., the Screening Procedures, Experimental Procedures, or Monitoring/Follow-up Procedures described above). However, the expenses for routine health check-ups or standard medical care for your medical problem (care you would have received whether or not you were in this study) is your responsibility (or the responsibility of your insurance provider or governmental program). You will be charged, in the standard manner, for any procedures performed for your standard medical care.

***What will happen if I am harmed as a result of taking part in this study?***

It is important that you report any suspected study-related illness or injury to the research team listed at the top of this form immediately. Compensation for an injury resulting from your participation in this research is not available from the University of Texas Southwestern Medical Center at Dallas or the Retina Foundation of the Southwest. You retain your legal rights during your participation in this research.

***Will my information be kept confidential?***

Information about you that is collected for this research study will remain confidential unless you give your permission to share it with others, or as described below.

You should know that the UT Southwestern Institutional Review Board may look at and/or copy your medical records. An Institutional Review Board (IRB) is a group of people who are responsible for assuring the community that the rights of participants in research are respected.

To help us further protect the information the investigators have obtained a Certificate of Confidentiality from the U.S. Department of Health and Human Services (DHHS). This Certificate adds special protections for research information that identifies you and will help researchers protect your privacy. This Certificate does not mean the government approves or disapproves of our project.

With this Certificate of Confidentiality, the researchers cannot be forced to disclose information that may identify you in any judicial, administrative, legislative, or other proceeding, whether at the federal, state, or local level. There are situations, however, that legally require disclosure, such as:

- to DHHS for audit or program evaluation purposes
- information regarding test results for certain communicable diseases to the Texas Department of State Health Services, including, but not limited to HIV, Hepatitis, Anthrax, and Smallpox
- if you pose imminent physical harm to yourself or others
- if you pose immediate mental or emotional injury to yourself
- if the researchers learn that a child has been, or may be, abused or neglected
- if the researchers learn that an elderly or disabled person has been, or is being, abused,

neglected or exploited .

The researchers will not, in any case, disclose information about you or your participation in this study unless it is included in the Authorization for Use and Disclosure of Protected Health Information for Research Purposes or it is required by law (as mentioned above).

The Certificate of Confidentiality does not prevent you or a member of your family from voluntarily releasing information about your involvement in this research study. In addition, the researchers may not use the Certificate to withhold information about your participation in this research study if you have provided written consent to anyone allowing the researchers to release such information (including your employer or an insurance company). This means that you or your family must also actively protect your privacy.

A Certificate of Confidentiality does not represent an endorsement of this research project by the Department of Health & Human Services or any other Federal government agency.

***Whom do I call if I have questions or problems?***

For questions about the study, contact Dr. Birch at 214-363-3911 during regular business hours. For urgent eye related medical concerns, contact Texas Retina Associates at 214-692-6941 after hours and on weekends and holidays.

For questions about your rights as a research participant, contact the UT Southwestern Institutional Review Board (IRB) Office at 214-648-3060.

**USE OF SAMPLES:** You may have preferences about how your samples are used. Please answer each question below by circling the appropriate answer (yes or no):

Yes	No	Do you agree that your remaining samples be kept for use in future research to learn about, prevent, or treat eye disorders?
Yes	No	Do you agree that your samples may be used for research to answer other medical questions that are not necessarily related to eye disorders?
Yes	No	Do you agree that one of the investigators (or someone else he/she chooses) may contact you in the future?
Yes	No	If a mutation is identified predisposing you to a genetic eye disease, do you wish to be informed?

**SIGNATURES: YOU WILL HAVE A COPY OF THIS CONSENT FORM TO KEEP.**

Your signature below certifies the following:

- You have read (or been read) the information provided above.
- You have received answers to all of your questions and have been told who to call if you have any more questions.
- You have freely decided to participate in this research.
- You understand that you are not giving up any of your legal rights.

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Participant's name (printed) and signature (if at least ten years of age) \_\_\_\_\_ Date \_\_\_\_\_ Time \_\_\_\_\_

Legally authorized representative's name (printed), signature, and relationship to the participant \_\_\_\_\_ Date \_\_\_\_\_ Time \_\_\_\_\_ AM/PM

**ASSENT OF A MINOR:** I have discussed by participation in this research with my mother or father or legal guardian and my study doctor, and I agree to participate.

Signature (participants from 10 to 18 years old) \_\_\_\_\_ Date \_\_\_\_\_ Time \_\_\_\_\_ AM/PM

**\*\*\*\* PLEASE DO NOT WRITE BELOW THIS LINE \*\*\*\***

Name (printed) and signature of person obtaining consent \_\_\_\_\_ Date \_\_\_\_\_ Time \_\_\_\_\_ AM/PM

Witness'/translator's name (printed) and signature \_\_\_\_\_ Date \_\_\_\_\_ Time \_\_\_\_\_ AM/PM

**INTERPRETER STATEMENT:**

I have interpreted this consent form into a language understandable to the participant and the participant has agreed to participate as indicated by their signature above.

Name of Interpreter (printed) \_\_\_\_\_ Signature of Interpreter \_\_\_\_\_ Date \_\_\_\_\_ Time \_\_\_\_\_ AM/PM