Title of Study: Retinal Pathophysiology in Infants and Adults

FOR YOUR INFORMATION ONLY - DO NOT SIGN

Consent to be part of a Research Study
To be conducted at
The University of Texas Southwestern Medical Center
Retina Foundation of the Southwest

Key Information about this Study

The purpose of this study is to compare the visual function with genetic mutation in patients with hereditary eye disease. The information we learn by doing this study may help us to develop some target treatments for inherited retinal degenerations.

Participants in this study will have a saliva sample collected. Visual function tests will be state-of-the-art and include retinal imaging, electrophysiology, visual fields, and night vision testing.

The genetic analysis of the saliva sample will be done in the laboratory after collection of the sample. Your participation is complete once the visual function test results have been reviewed, and the saliva sample has been collected.

There is a risk of a scratch from a contact lens used in testing. Risks of taking the blood sample are discomfort and/or bruising; infection, excess bleeding, clotting, or fainting are also possible.

If you are interested in learning more about this study, please continue to read below.

Information about this form

Enrolling Children
If you are providing consent for someone else, for example your child, your next-of-kin or someone for whom you are the legal guardian or are designated as a surrogate decision maker on a medical power of attorney, please note that in the sections that follow, the word “you” refers to the person you are providing consent for.

You may be eligible to take part in a research study. This form gives you important information about the study.

Please take time to review this information carefully. You should talk to the researchers about the study and ask them any questions you have. You may also wish to talk to others (for example, your friends, family, or a doctor) about your participation in this study. If you decide to take part in the study, you will be asked to sign this form. Before you sign this form, be sure you understand what the study is about, including the risks and possible benefits to you.

Please tell the researchers or study staff if you are taking part in another research study.

Voluntary Participation - You do not have to participate if you don't want to. You may also leave the study at any time. If you decide to stop taking part in this research study, it will not affect your relationship with the UT Southwestern staff, the Retina Foundation of the Southwest staff, or doctors. Whether you participate or not will have no effect on your legal rights or the quality of your health care.

If you are a medical student, fellow, faculty, or staff at the Medical Center, your status will not be affected in any way.

General Information – “Who is conducting this research?”

Principal Investigator
The Principal Investigator (PI) is the researcher directing this study; the PI is responsible for protecting your rights, safety and welfare as a participant in the research. The PI for this study is David G. Birch, Ph.D., Department of Ophthalmology at UT Southwestern and of the Retina Foundation of the Southwest.

In the future, it is possible that the results of this research could result in a financial benefit to the Retina Foundation of the Southwest and/or the principal investigator. This institution has taken steps to not let this interfere with the way the study is conducted or your safety.

If you require further information regarding the financial arrangements described in this paragraph, you should discuss the matter with the Principal Investigator.

Funding
Local philanthropic agencies, non-profit organizations that promote scientific research, are funding this study. These organizations are providing money to the Retina Foundation of the Southwest so that the researchers can conduct the study.

Purpose – "Why is this study being done?"

You are asked to participate in this research study of inherited genetic retinal disorders to learn more about these conditions and to gather information from patients and their families on how their vision might change over time. The goal is to eventually find possible treatment and offer treatment trials to those that can qualify.

The researchers hope to learn which genetic mistakes (mutations) cause retinal disorders and what symptoms are seen with different types of mutations and how they change over time.

Information about Study Participants – "Who is participating in this research?"

You are being asked to be a participant 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.

This study will enroll approximately 400 study participants each year.

Information about Study Procedures – "What will be done if you decide to be in the research?"

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.

Study Procedures - as a participant, you will undergo the following procedures:

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.

Optical Coherence Tomography (OCT) and Optical Coherence Tomography Angiography (OCT-A: This procedure is non-harmful and should not cause you any discomfort.

High-Resolution Optical Coherence Tomography (HR-OCT: Heidelberg Engineering SPECTRALIS HR-OCT is an investigational device and is not FDA approved. It is considered to be Non-Significant Risk (NSR) device. HR-OCT is a non-contact ophthalmic diagnostic medical device that uses existing technology and does not present potential risk to your health. However, the bright lights during imaging might cause temporary discomfort.

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.

Your tissue contains DNA. DNA makes up the genes that serve as the "instruction book" for the cells in our bodies. By studying genes, researchers can learn more about diseases such as cancer. There are many different types of genetic tests. The testing on your tissue samples might include genetic testing called whole genome sequencing. Whole genome sequencing looks at all the known genes in your cells. This type of testing can provide useful information to researchers. It can also present risks if the test results became known to others, for example you could have problems with family members or insurance companies. There is also a risk that these test results could be combined with other genetic information to identify you.

It is possible that this study will identify information about you that was previously unknown, such as disease status or risk. Any clinically relevant results of the research will be communicated to you. Clinically relevant means that the information indicates that you may be at risk for a serious illness known at the time of testing to be treatable and it can be confirmed. In that case, we will attempt to notify you using the contact information you have provided. Genetic counseling is available through Kaylie Jones at the Retina Foundation of the Southwest.

If you do not want to be notified of any of these incidental findings, please initial below.

_____ Please do not notify me of any incidental findings obtained from this research.

Risks – "What are the risks of participation in the research?"

Risks from the research

Risks from the specific research procedures (drug(s), interventions, or procedures)
There are risks to taking part in this research 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 your 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.

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

Genetic Informational risks
This research study includes genetic testing. Human tissue contains genes that determine many of a person's physical characteristics, such as the color of eyes and hair. In some cases, genetic testing of tissues can be used to indicate a risk for the development of certain diseases. Genetic information is unique to each individual and could potentially be used to discover possible changes in a person's future health status or life expectancy, or that of his/her children and family members.

Releasing this information to you could cause psychological distress, anxiety or family problems. Releasing this information to others, such as including it in your medical record, may pose a possible risk of discrimination, or increase difficulty in obtaining or maintaining disability, long-term care, or life insurance.

These risks would occur if your information is released by mistake. The measures being taken to protect your privacy are discussed below and make this possibility unlikely.

Even though the results of genetic testing may not be linked to you, it is possible that people of your ethnic background may be found to be at more risk for certain diseases based on future genetic research and this information might harm you in the future as a member of the group. Also, there may be unknown risks of genetic testing in the future.

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?"

Risks and side effects related to the dilating drops used include those which are:

Rare and Serious
In 100 people, approximately 1 or less may have a sudden increase in intraocular pressure (IOP):

For more information about risks and side effects, ask one of the researchers or study staff.

Are there Risks related to withdrawing from the study?
No, you may choose to withdraw participation at any time without risk.

What if a research-related injury occurs?
The researchers have taken steps to minimize the known or expected risks. However, you may still experience problems or side effects, even though the researchers are careful to avoid them. In the event of a research-related injury or if you experience an adverse reaction, please immediately contact your study doctor. See the section "Contact Information" for phone numbers and additional information. You may also need to tell your regular doctors.

If you have an injury or illness from the procedures required for this study, medical care will be provided. Depending on the circumstances, the reasonable medical expenses required to treat such injury or illness may be paid for by the study sponsor.

The coverage for such injury or illness is only available if the principal investigator and study sponsor, if applicable, have decided that the injury/illness is directly related to the study procedures and is not the result of a pre-existing condition or the normal progression of your disease, or because you have not followed the directions of the study doctor. If your insurance is billed, you may be required to pay deductibles and co-payments that apply. You should check with your insurance company about any such payments.

We have no plans to give you money if you are injured. If you sign this form, you do not give up your right to seek additional compensation if you are harmed as a result of being in this study.

Benefits – "How could you or others benefit from your taking part in this study?"
You may not receive any personal benefits from being in this study.

We hope the information learned from this study will benefit other people with similar conditions in the future.

Costs – Will taking part in this study cost anything?

You or your health insurance company will be responsible for the cost of treatments and procedures that would be done whether or not you took part in this study, such as routine health check-ups or standard medical care for your medical problem. It is important to understand that some insurance companies do not cover some costs (for example, approved drugs used in a way different from the package instructions). If your insurance company does not cover these treatments or procedures, you will be required to pay for them.

Ask the researchers if you have any questions about what it will cost you to take part in this study (for example bills, fees, or other costs related to the research). All tests and procedures conducted at the Retina Foundation of the Southwest will be free of charge to you and will not be filed with your insurance. If you are eligible, your DNA can be sent to a research lab to attempt to identify your genetic mutation that is causing your eye problem. The result from this type of lab can take some time. If you wish to get answers quickly, there are commercial labs available, but you would be responsible for the cost of that test.

Confidentiality – How will your records be kept confidential?

Information we learn about you in this study will be handled in a confidential manner, within the limits of the law. If we publish the results of the study in a scientific journal or book, we will not identify you. The Institutional Review Board and other groups that have the responsibility of monitoring research may want to see study records which identify you as a subject in this study.

Certificate of Confidentiality:
To help us further protect your information, the investigators will obtain 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.

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, where we will voluntarily disclose information consistent with state or other laws, 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; or
- if the researchers learn that an elderly or disabled person has been, or is being, abused, neglected or exploited.

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.

How will my information and/or tissue samples be used?
With appropriate permissions, your samples and collected information may also be shared with other researchers here, around the world, and with companies.

By agreeing to participate in this study, your information or tissue samples could be used for future research studies or sent to other investigators for future research studies without additional consent from you. The information that identifies you will first be removed from your information or tissue samples. If you do not want your information or tissue samples to be used for future research studies without your consent, you should not participate in this study.

Research policies require that private information about you be protected and this is especially true for your health information. However, the law sometimes allows or requires others to see your information. The information given below describes how your privacy and the confidentiality of your research records will be protected in this study. Medical information collected during this study and the results of any test or procedure that may affect your medical care may be included in your medical record. The information included in your medical record will be available to health care providers and authorized persons including your insurance company.

What is Protected Health Information (PHI)?
Protected Health Information is information about a person's health that includes information that would make it possible to figure out whose it is. According to the law, you have the right to decide who can see your protected health information. If you choose to take part in this study, you will be giving your permission to the investigators and the research study staff (individuals carrying out the study) to see and use your health information for this research study. In carrying out this research, the health information we may see and use about you will include medical and treatment history and eye records from a referring eye doctor.

How will your PHI be shared?
Because this is a research study, we will be unable to keep your PHI completely confidential. We may share your health information with people and groups involved in overseeing this research study including:

- the members of the local research team.
- The Institutional Review Board, Human Research Protection Program Office and the Compliance Office of the University of Texas Southwestern Medical Center, and other groups that oversee how research studies are carried out.
- The Research offices at the University of Texas Southwestern Medical Center,.
- Representatives of domestic and foreign governmental and regulatory agencies may be granted direct access to your health information for oversight, compliance activities, and determination of approval for new medicines, devices, or procedures.

If you decide to participate in this study, you will be giving your permission for the groups named above, to collect, use and share your health information. If you choose not to let these groups collect, use and share your health information as explained above, you will not be able to participate in the research study.

Parts of your PHI may be photocopied and sent to a central location or it may be transmitted electronically, such as by e-mail or fax. The groups receiving your health information may not be obligated to keep it private. They may pass information on to other groups or individuals not named here.

The Genetic Information Nondiscrimination Act (GINA) is a Federal law that will protect you in the following ways:

- Health insurance companies and group plans may not request genetic information from this research;
- Health insurance companies and group plans may not use your genetic information when making decisions regarding your eligibility or premiums;
- Employers with 15 or more employees may not use your genetic information when making a decision to hire, promote, or fire you or when setting the terms of your employment.

GINA does not protect you against genetic discrimination by companies that sell life insurance, disability insurance, or long-term care insurance. GINA also does not protect you against discrimination based on an already-diagnosed genetic condition or disease.

How will your PHI be protected?
In an effort to protect your privacy, the study staff will use code numbers instead of your name, to identify your health information. Initials and numbers will be used on any photocopies of your study records, and other study materials containing health information that are sent outside of the Retina Foundation of the Southwest for review or testing. If the results of this study are reported in medical journals or at meetings, you will not be identified.

Do you have to allow the use of your health information?
You do not have to allow (authorize) the researchers and other groups to see and share your health information. If you choose not to let the researchers and other groups use your health information, there will be no penalties but you will not be allowed to participate in the study.

After you enroll in this study, you may ask the researchers to stop using your health information at any time . However, you need to say this in writing and send your letter to Dr. David G. Birch at the Retina Foundation of the Southwest, 9600 N Central Expressway, Suite 200. Dallas TX 75231. If you tell the researchers to stop using your health information, your participation in the study will end and the study staff will stop collecting new health information from you and about you for this study. However, the study staff will continue to use the health information collected up to the time they receive your letter asking them to stop.

Can you ask to see the PHI that is collected about you for this study?
The federal rules say that you can see the health information that we collect about you and use in this study. Contact the study staff if you have a need to review your PHI collected for this study.

FOR YOUR INFORMATION ONLY - DO NOT SIGN

How long will your PHI be used?
By signing this form, you agree to let us use and disclose your health information for purposes of the study until the end of the study. This permission to use your personal health information expires when the research ends and all required study monitoring is over.

Contact Information – Who can you contact if you have questions, concerns, comments or complaints?

If you have questions now, feel free to ask us. If you have additional questions, concerns, comments or complaints later or you wish to report a problem which may be related to this study please contact: Dr. David G. Birch PhD., Kirsten G. Locke, C.R.A. or Martin Klein, M.S.

Primary contact:
David G. Birch, Ph.D. can be reached at 214-363-3911
If primary is not available, contact
Kirsten G. Locke or Martin Klein can be reached at 214-363-3911

The University of Texas Southwestern Medical Center Human Research Protection Program (HRPP) oversees research on human subjects. HRPP and Institutional Review Board (IRB) representatives will answer any questions about your rights as a research subject, and take any concerns, comments or complaints you may wish to offer. You can contact the HRPP by calling the 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 you 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?

Research Consent & Authorization Signature Section

If you agree to participate in this research and agree to the use of your protected health information in this research, sign this section. You will be given a copy of this form to keep. You do not waive any of your legal rights by signing this form.

SIGN THIS FORM ONLY IF THE FOLLOWING STATEMENTS ARE TRUE:

- You have read (or been read) the information provided above.
- Your questions have been answered to your satisfaction about the research and about the collection, use and sharing of your protected health information.
- You have freely decided to participate in this research, or you are voluntarily giving your consent for another person to participate in this study because you believe this person would want to take part if able to make the decision and you believe it is in this person's best interest.
- You understand that a copy of this signed consent document, information about this study, and the results of any test or procedure that may affect your medical care, may be included in your medical record. Information in your medical record will be available to health care providers and authorized persons including your insurance company.
- You authorize the collection, use and sharing of your protected health information (another person's protected health information) as described in this form.

Adult Signature Section

FOR YOUR INFORMATION ONLY - DO NOT SIGN

Printed Name of Participant	Signature of Participant	Date	Time AM PM
Printed Name of Person Obtaining Consent	Signature of Person Obtaining Consent	Date	Time AM PM

Surrogate Signature Section

FOR YOUR INFORMATION ONLY - DO NOT SIGN

Printed Name of Participant	Signature of Participant Giving Assent (*If incapable of signing, person obtaining consent should initial here*)	Date	Time AM PM
Printed Name of Person Giving Consent for Participant (If applicable)	Signature of Person Giving Consent ☐Parent/☐Guardian/☐Legally Authorized Representative	Date	Time AM PM
Printed Name of Person Obtaining Consent	Signature of Person Obtaining Consent	Date	Time

Witness / Interpreter Signature Section

Interpreter/witness (Interpreter signature required per hospital policies when physically present.)

I attest that I have interpreted the information in this consent form and it was explained to, and apparently understood by the subject or the subject's legal authorized representative, and that informed consent was freely given by the subject or the subject's legally authorized representative as indicated by their signature on the associated **short form**.

Printed Name of Interpreter	Signature of Interpreter	Date	Time AM PM

FOR YOUR INFORMATION ONLY - DO NOT SIGN

Witness Signature (required when interpreter is not physically present-e.g., Language Line is used):

By signing below:

I attest that the information in the consent form was accurately explained to, and apparently understood by the subject or the subject's legal authorized representative, and that informed consent was freely given by the subject or the subject's legally authorized representative as indicated by their signature on the associated **short form**.

Printed Name of witness	Signature of witness	Date	Time AM PM

Blind or Illiterate Signature Section *At the time of consent, also complete this section if consent is obtained from an individual who is unable to read and/or write but can otherwise communicate and/or comprehend English (e.g., blind, physically unable to write, etc.)*

Declaration of witness:

By signing below, I confirm I was present for the entire consent process. The method used for communication (e.g., verbal, written, etc.) with the subject was: _____ .

The specific means (e.g., verbal, written, etc.) by which the subject communicated agreement to participate was: _____.

Printed Name of Witness	Signature of Witness	Date	Time AM PM