REQUEST FOR MOLECULAR STUDIES FORM

GENETICS OF INHERITED RETINAL DISORDERS

DIVISION OF HUMAN GENETICS, WERNHER & BEIT NORTH
FACULTY OF HEALTH SCIENCES, UNIVERSITY OF CAPE TOWN, OBSERVATORY, 7925
TEL: 021 406-6995 EMAIL: retina@uct.ac.za

PATIENT DETAILS

SURNAME: _____ **NAME:** _____

DATE OF BIRTH: ____/ ____/_____ **SEX: FEMALE - ☐ MALE - ☐** **ETHNICITY:**_____

NEW FAMILY: YES - ☐ NO - ☐ (If NO please fill in family name) FAMILY NAME: _____

Number of Children: _____ **Number of affected family members:** _____

CONTACT ADDRESS: _____

_____**CODE** _____

TEL: …….. **E-mail** ……

REFERRAL SOURCE

PLEASE NOTE: A confirmation of diagnosis (COD) form is required to accompany all samples. This separate form needs to be completed by an Ophthalmologist and can be sent separately.

NAME OF REFERRING DOCTOR: _____ REFERRING FACILITY: _____

TEL: _____ E-mail: _____

ADDRESS: _____

REASON FOR REFERRAL (CLINICAL DIAGNOSIS)

AFFECTED - ☐ AT RISK - ☐ CARRIER - ☐ SPOUSE - ☐ UNAFFECTED - ☐

RETINITIS PIGMENTOSA ☐	USHER SYNDROME ☐	**DOMINANT INHERITANCE ☐**
STARGARDT DISEASE ☐	MACULAR DYSTROPHY ☐	**RECESSIVE INHERITANCE ☐**
ARMD – WET ☐	ARMD – DRY ☐	**X-LINKED INHERITANCE ☐**
OTHER DISORDER:	**AGE OF ONSET:**	**ISOLATED CASE ☐**
	DIAGNOSIS AGE:	

FAMILY HISTORY INFORMATION

ADDITIONAL FAMILY HISTORY _____

ADDITIONAL DISORDERS (APPARENT OR PREVIOUSLY TREATED):_____

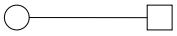
RELEVANT CLINICAL DETAILS: _____

PHYSICAL DISABILITY-☐ INTELLECTUAL DISABILITY- ☐ DEAFNESS - ☐ IMPAIRED VISION - ☐
NIGHT BLINDNESS - ☐ AGE OF ONSET: _____ OTHER: _____

PEDIGREE/ FAMILY TREE (If more extensive, please use a separate page)

Maternal Ethnicity/ Genetic Origin: _____ Paternal Ethnicity/ Genetic Origin: _____

Grandparents

Parents/ Siblings 

Proband (Patient)/ Siblings

Children

Have samples from this patient been sent to a DNA lab before? Yes - ☐ No - ☐ Don't Know - ☐.
If "Yes": Where: _____ When: _____

Have samples from other family members previously been sent to a DNA lab for genetic ophthalmic disease testing?
Yes - ☐ No - ☐ Unsure - ☐.

If "Yes": Name of lab: _____ Mutation identified: _____

RETINA SA MEMBERSHIP

This is not a requirement, but is necessary information to assist us with our database.

RETINA SOUTH AFRICA
a Cure in Sight for Blindness

Are you currently a Retina SA member? Yes - ☐ No - ☐

If "No", have you ever been a Retina SA member? Yes - ☐ No - ☐

TEST REQUESTED

ABCA4 Quick 7 - ☐ UCT IRD panel - ☐ Overseas test" - ☐ Specify: _____

Known family mutation - ☐ Specify: _____ Other - ☐ Specify: _____

For Laboratory use only: DNA number: _____ Volume Saliva OR Blood: _____ (ml)

Date Received: Year: _____ Month: _____ Day: _____ Computer Index No: _____

SPECIMEN TUBES REQUIRED: EITHER 1X Oragene saliva kit OR 2X 4 ml Plastic purple top tubes (containing EDTA). Name and Date of birth to be written on each tube.

BLOODS ARE TO BE KEPT REFRIGERATED, CAREFULLY PACKAGED AND TRANSPORTED IN A POLYSTYRENE COOLBOX WITH AN ICE BRICK - DO NOT FREEZE. SALIVAS ARE TRANSPORTED AT ROOM TEMPERATURE. SAMPLES ARE CODED ON ARRIVAL IN THE LABORATORY ACCORDING TO THE FAMILY NAME. WHEN AVAILABLE, RESULTS ARE GIVEN TO PARTICIPANTS.

GENETICS OF INHERITED RETINAL DISORDERS
CONSENT FORM REQUIRED FOR DNA ANALYSIS AND STORAGE

1. I, _____ (*NAME*), request that an attempt be made using genetic material to assess the probability that, I / my child / my unborn child, might have inherited a disease-causing mutation in the gene for:

_____*(NAME OF DISORDER)*

2. No portion of the sample can be stored for later use ☐ *(MARK IF APPLICABLE)*

OR

The data and a portion of the sample can be stored indefinitely for:

a. Possible re-analysis
b. Analysis for the benefit of members of my immediate family

(*DELETE WHERE NOT APPLICABLE)*

3. Only meaningful results that have clear diagnostic implications will be given to me, via my doctor or genetic counsellor: _____ (*NAME*) if and when available. In addition, these results can be made known to my family member / friend:

_____ (*NAME*)

4. My doctor(s) will provide relevant clinical details to the Division of Human Genetics, UCT.

5. Confidential information may need to be disclosed if there is clear evidence that someone is at risk of harm or abuse.

6. I may be contacted about this genetic research or future research relevant to me (e.g. treatment trials) in the future. I may withdraw from such contact at any time.

7. I may withdraw my consent and/or biological material at any time, without this affecting my future medical care.

I have explained genetic testing (including the risks, benefits, and alternatives) to this individual. I have addressed the limitations and I have answered this person's questions to the best of my ability in a language that they understand.
This consent form was signed in my presence. _____

(Please print recruiter/consenter name clearly)

DATE _____ /_____ /_____ **PLACE:** _____

PATIENT SIGNATURE: _____ **WITNESS CONSENT:** _____

The Division of Human Genetics at the University of Cape Town (UCT) has a long-term research project, studying the genetics of inherited retinal diseases. You have been asked to participate because you (or a family member) have one of the retinal diseases we study. Thousands of people are already taking part in our study, which began in 1985. Although you have been invited to join the study, you may refuse at any time, without it affecting the care you receive from your doctor.

If you decide to join our study, we will collect basic information from you, and your doctor will provide us with clinical information about your retinal disease. These records will be securely stored in paper form and electronically in our offices at UCT. You will donate a sample of saliva or blood and we will extract your genetic material (DNA) from it. The DNA will be given an anonymous code and stored in our repository (freezers). The only people who will be able to match your personal details to the DNA code are researchers and genetic counsellors in the Division of Human Genetics, who have all signed an oath of confidentiality. Collaborations with international facilities may require that samples and clinical information be shared. In these cases, only the code will be shared so we maintain full patient confidentiality.

The sample may also be used for future research into inherited retinal diseases that is approved by the UCT Human Research Ethics Committee, provided that any information from such research will remain confidential.

The big focus of our study is to understand the genetics of inherited vision loss. We try to find the change in the gene which cause vision loss. If we find the change in the gene responsible in your family, we will write you a report which a genetic counsellor or your doctor can explain to you. This may be helpful because it can give you the risks for your family members. Your doctor may find the genetic information useful, and it also may be helpful to know if a therapy is ever developed for that gene.

- Many of the risks associated with genetic testing involve the emotional, social, or financial consequences of the test results. People may feel angry, depressed, anxious, or guilty about their results.
- Genetic testing could create tension within a family because the results can reveal information about other family members in addition to the person who is tested.

- The possibility of genetic discrimination in employment or insurance is also a concern.
- Genetic testing can provide only limited information about an inherited condition. The test often can't determine if a person will show symptoms of a disorder, how severe the symptoms will be, or whether the disorder will progress over time.
- Another major limitation is the lack of treatment strategies for many genetic disorders once they are diagnosed.
- The analysis procedure is specific to the genetic condition related to the visual impairment mentioned above and cannot determine the complete genetic makeup of an individual.
- Genetic analysis may not be informative for some families or family members.
- Even under the best conditions, current technology of this type is not perfect and could lead to incorrect results.
- Where biological material is used for research purposes, there may be no direct benefit to you.
- The genetics laboratory is under an obligation to respect medical confidentiality, but confidential information may need to be disclosed if there is clear evidence that someone is at risk of harm or abuse.

Your samples and information will only be used to study inherited retinal diseases, not any other diseases. The samples and information will remain stored in the Division and will carry on being used unless you contact us and ask us to destroy them (which you can do at any time, with no risk to the level of care you will receive).

The UCT Faculty of Health Sciences Human Research Ethics committee can be contacted on 021 4066338 in case you have any ethical concerns or questions about your rights or welfare as a participant on this research study.

For any questions regarding the ethics governing the use of this repository, please contact:

The Head of the Human Research Ethics Committee; **Prof Marc Blockman**

Marc.Blockman@uct.ac.za and/or

Custodian of this repository; **Prof Raj Ramesar** **Raj.Ramesar@uct.ac.za** or **retina@uct.ac.za**

Tel: 021 4066297