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 FAX: 021 650-2010 EMAIL: lisa.roberts@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: …….. FAX: …… 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 faxed separately.

NAME OF REFERRING DOCTOR: _____ REFERRING FACILITY: _____

FAX: _____ 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 - ☐ "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 2 X 4 ml Plastic purple top tubes (containing EDTA). NAME AND DATE OF BIRTH TO BE 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 ACCORDING TO AN ESTABLISHED PROTOCOL

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

1. I, _____, 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. I understand that the genetic material for analysis is to be obtained from: blood cells / skin sample/ other (specify)

3. I request that no portion of the sample be stored for later use. ☐ *(MARK IF APPLICABLE)*

OR

I request that the data and a portion of the sample be stored indefinitely for:

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

(*DELETE WHERE NOT APPLICABLE)*

4.The sample may also be used for

- Future research purposes, not yet identified, subject to the approval of the University of Cape Town Human Research Ethics Committee, provided that any information from such research will remain confidential.
- Although samples are stored in the Division of Human Genetics, collaborations with international facilities may require that samples and clinical information be shared. In these cases, all samples will be de-identified and given a unique code, and full patient confidentiality will be maintained.

5. Regarding the results of any current and future analysis carried out on this sample of stored biological material: only meaningful results that have clear diagnostic implications will be made known to me, via my doctor, in accordance with the relevant protocol, if and when available. In addition, I authorise that these results may be made known to: *(DELETE WHERE NOT APPLICABLE)*

Doctor / Family member / Friend: _____

_____

6. I authorise / do not authorise my doctor(s) to provide relevant clinical details to the Division of Human Genetics, UCT.

7. I have been informed that:

- 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 me.
- The genetics laboratory is under an obligation to respect medical confidentiality.

8. Please note that confidential information may need to be disclosed if there is clear evidence that someone is at risk of harm or abuse.

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

10. Ethical Approval: Written approval has been granted by the University of Cape Town Ethics Committee (HREC).
For any questions regarding the ethics of this repository, please contact:
Prof Marc Blockman- **Marc.Blockman@uct.ac.za** and/or
Custodian of this repository-Prof Raj Ramesar
Raj.Ramesar@uct.ac.za **- 021 406-6297**

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 name clearly)

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

PATIENT'S SIGNATURE: _____ **WITNESS CONSENT:** _____