Department of Ophthalmology
Faculty of Medicine and Dentistry

2319, 10240 Kingsway Avenue
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Tel: (780) 735-5954
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INFORMATION SHEET AND CONSENT

Title of Research Study **Choroideremia: Expanding the Phenotype and Genotype.**

Principal Investigator: Dr Ian MacDonald, U Alberta and Alberta Health Services
780-735-5932

Background:

Our goal is to develop therapies for choroideremia (CHM), a hereditary cause of vision loss. We want to learn more about the genetic cause of CHM by continuing to analyze CHM patients and also re-examining patients in whom no mutation was previously found.

A survey of CHM patients' general health beyond their eye health has not been published. CHM has been considered to be an eye problem, with limited attention paid to other health problems unrelated to the eye. We would like your permission to allow us to review your ocular (eye) health, general health and history of drug use. This will allow us to discover other health effects of CHM that have not yet been considered, and also any potential drug toxicity concerns that are important for CHM patients.

Recent work from our laboratory and those of our colleagues at the National Eye Institute of the NIH has shown that certain proteins are released into the blood at different levels in stages of the disease. We would like to follow-up on these observations by including a larger sample of patients.

Purpose:

We are requesting your permission to review your health records to find common issues in ocular health, general health and reactions to drugs amongst CHM patients. We are also interested in studying the potential toxicity concerns of drugs in CHM patients This will allow us to discover other health effects of CHM that have not yet been considered.

If you agree, we would like to obtain a blood sample or a small piece of skin to grow cells in our laboratory. The blood will be used to identify and study the genes and proteins associated with retinal degeneration in your family. We may create a cell line from your blood cells that will allow us to obtain additional material (protein, RNA and DNA) to study in the future without having to draw more blood. Research on your cells may lead us to recommend safer drug therapies for CHM patients.

Procedures: Participating in this study will allow us to review your health records. If you agree to participate, we will record your visual acuity, visual field, and the function and images of your retina. These are not difficult procedures and are part of the routine care of patients. Please initial one or both of the two lines below if you want to consent to these procedures. Leave them blank if you do not consent to them.

_____We will also ask some patients for permission to draw blood (approximately 15 ml or 3 tbsp). The sample will be used for genetic analysis and protein analysis to help confirm your clinical diagnosis of CHM and monitor its progress.

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_____ If you agree we will also do a skin biopsy. If you are allergic to any medications you must inform us. After numbing the skin with a local anaesthetic we will remove a piece of skin measuring 2 mm, usually taken from your inner arm, using a small instrument. A small dressing will be applied to the biopsy area and kept in place for 24 hours. You will be given instructions for care of the biopsy site to take home, along with some ointment which you need to apply.

The blood, cell lines, protein, RNA and DNA samples will be coded so that participant identification is only possible with a key. Samples will be stored in secured freezers on the University of Alberta campus or outside laboratories that we select for this project. The key to the code will be kept in a separate, secure area. These samples will be used for the study described in these information sheets.

If a future research project arises where your samples could be useful, we ask for your decision whether or not your sample can be used. Please initial by the line that you agree to:

I give permission to use my blood of other fluids, tissues or samples in future research studies under the following conditions: (initial only the conditions you consent to)

_____ These samples may be used for any research project that has approval from the governing research ethics board. It does not matter to me if the sample is identifiable as mine. You do not need to contact me if my sample is used for another study.

_____ These samples may be used for future studies without contacting me if all identifying information is removed so that the sample cannot be identified as mine.

_____ I wish to be contacted if further studies with my samples are considered. After the study has been explained, I will decide if I want my samples to be included.

_____ Under no circumstances shall the samples be used for future studies. Samples should be discarded once the present study is complete.

If future research on your sample provides information important about your health, we will try to contact you. If you wish to be contacted, please keep the principal investigator for this study updated about changes in your address or phone number.

If you withdraw from this research project before it is complete, any samples you have contributed will be discarded at the point of your withdrawal. Results obtained prior to your withdrawal from the study will be maintained and your privacy will be preserved.

Possible Benefits: The possible benefits to you for participating in this study are that you may contribute to a greater understanding of CHM and how it affects both the eyes and the rest of the body. This knowledge may uncover possible drug toxicity issues for CHM patients which we can then alert you of, and hopefully recommend safer drug therapies for you.

Possible Risks: There may be bruising at the site of the blood draw if you agree to provide a sample. There is a small risk of fainting or infection in the area of the blood draw.

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There may be a small scar in the area where the skin biopsy sample is obtained. Risks of the biopsy, although rare, include bleeding, pain, infection, or reaction to the anesthetic. If you have allergies, you must inform us.

Confidentiality: Personal health records relating to this study will be kept confidential. Any research data collected about you during this study will not identify you by name, only by your initials and a coded number. Your name will not be disclosed outside the clinic. Any report published as a result of this study will not identify you by name.

For this study, the doctor will want to access your personal health records for health information such as past medical history and prescribed drugs. He may also need to contact your family physician and your other health care providers to obtain additional medical information. The health information collected as part of this study will be kept confidential unless release is required by law, and will be used only for the purpose of the research study. By signing the consent form you give permission to the study staff to access any personally identifiable health information which is under the custody of other health care professionals as deemed necessary for the conduct of the research.

By signing the consent form you give permission for the collection, use and disclosure of your medical records. In Canada, study information is required to be kept for 25 years. Even if you withdraw from the study, the medical information which is obtained from you for study purposes will not be destroyed. You have a right to check your health records and request changes if your personal information is incorrect.

Voluntary Participation: You are free to refuse to participate in this study, and/or refuse to provide a blood sample or skin biopsy, and your continuing medical care will not be affected in any way. If the study is not undertaken or if it is discontinued at any time, the quality of your medical care will not be affected.

Contact Names and Telephone Numbers:
If you have concerns about this study, you may contact the Research Ethics Office at 780-492-2615. This office has no direct involvement with this project.

Please contact the individual identified below if you have any questions or concerns:

Dr Ian MacDonald, Chair and Chief, Department of Ophthalmology 780-735-5932

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Title of Project: Choroideremia: Expanding the phenotype and genotype

Principal Investigator: Dr Ian MacDonald **Phone Number: 780-735-5954**

Adult Patient's Consent:

	Yes	No
Do you understand that you have been asked to be in a research study?	☐	☐
Have you read and received a copy of the Information Sheet?	☐	☐
Do you understand the benefits and risks involved in taking part in this research study?	☐	☐
Have you had an opportunity to ask questions and discuss this study?	☐	☐
Do you understand that you are free refuse to participate, without having to give a reason and without affecting your future medical care?	☐	☐
Has the issue of confidentiality been explained to you?	☐	☐
Do you understand who will have access to your records, including personally identifiable health information?	☐	☐
Do you want the investigator(s) to inform your family doctor that you are participating in this research study? If so, give his/her name _____	☐	☐

Who explained this study to you?

I agree to take part in this study: YES ☐ NO ☐

Signature of Research Subject _____

(Printed Name) _____Date:_____

I believe that the person signing this form understands what is involved in the study and voluntarily agrees to participate.

Signature of Investigator or Designee _____ Date _____

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Parent's Permission for Minor Patient
I have read the information sheet for this study and have had opportunities to discuss it and ask questions. I know that I may ask more questions, and that my child may ask questions as well.
I give permission for my child to take part in this study.
I know that I may change my mind and withdraw my child without offering a reason.

_____
Signature of Parent(s)/Guardian(s) and Date

Child's Verbal Assent (if applicable)
The information in the above consent was described to my child and my child agrees to participate in this study.

_____
Signature of Parent(s)/Guardian(s) and Date

_____
Signature of Study Staff and Date