Centre universitaire de santé McGill | McGill University Health Centre

INFORMATION AND CONSENT FORM

Registry and Biobank Participation

Adult Group

Name of the biobank:	SCCOHT/SMARCA4 Registry and Biobank
MUHC REB number:	2021-6537
Responsible researcher:	Dr. William D. Foulkes, MBBS, PhD, FRCPC Professor, Departments of Medicine, Oncology and Human Genetics, McGill University
Sites:	Research Institute of the McGill University Health Centre
Support:	Department of Defense Congressionally Directed Rare Cancers Research Program, USA The Eve Appeal, UK Small Cell Ovarian Cancer Foundation, USA Research Institute of the MUHC, Canada

For the purpose of this research information and consent form, "you" will refer to the person diagnosed with small cell carcinoma of the ovary, hypercalcemic type (SCCOHT) and/or with an inherited variant in the *SMARCA4* gene. The person entering the information may be the eligible individual or a family member or guardian of the eligible individual (the person legally responsible for the care and maintenance of the eligible individual) or a relative of a deceased individual.

INTRODUCTION

We invite you to take part in the SCCOHT/SMARCA4 Registry and Biobank (the "Registry/Biobank") because you have been diagnosed with SCCOHT and/or have an inherited variant in the *SMARCA4* gene. This consent form describes the Registry/Biobank and what it means to participate. Before you accept to take part in this project and sign this information and consent form, please take the time to read, understand and carefully examine the following information. You may also want to discuss participation in this project with your family doctor, a family member and/or a close friend.

We invite you to speak to the researcher responsible for the Registry/Biobank or to other members of the research team and ask them any questions you may have about this project. Please also ask a member of the research team about any parts of this consent form that you do not understand. You are also encouraged to seek assistance from your treating physician to fill out medically relevant sections or to provide requested medical documents as needed. All participation is voluntary, and you are not under any obligation to participate.

BACKGROUND AND OVERVIEW

A research registry is a place where medical information, family history and other related information from participants is collected and stored for future research. We would like to invite you to provide medical information on your disease and/or diagnosis for the Registry. We will ask you to complete medical questionnaires and seek your permission to review your medical records. This registry is linked to a biobank, which is a place that stores tissue, blood or other samples from participants and provides them to scientists for future research. We will also invite you to have some of your biological samples (e.g. blood) stored in the Biobank. We will try to get biological material during your planned clinical procedures or from leftover tissue after such procedures (e.g. request to the pathology department).

Small cell carcinoma of the ovary, hypercalcemic type (SCCOHT) is a rare and aggressive type of ovarian cancer, which mainly affects young women. Inherited variants in the *SMARCA4* gene have been identified in all studied familial cases of SCCOHT, however no therapies targeting these variants are yet available. The rarity of SCCOHT makes it difficult to study and therefore difficult to manage and treat.

Inherited *SMARCA4* variants may be associated with SCCOHT, but in some people, especially men (as they do not have ovaries) there are no obvious ill-effects. In other cases, variants in *SMARCA4* have been associated with Coffin-Siris syndrome, a very rare genetic disorder characterized by developmental delays that is typically diagnosed early on in life. We do not know why some *SMARCA4* variants have no effect or why some cause Coffin-Siris syndrome, while others cause SCCOHT and related cancers. This Registry/Biobank hopes to help researchers find answers to these questions.

PURPOSE OF THE REGISTRY/BIOBANK

The SCCOHT/SMARCA4 Registry and Biobank was created to be a comprehensive database that includes individuals with a SCCOHT diagnosis as well as individuals with inherited variants in the *SMARCA4* gene from across the world. More specifically, the goals of this Registry/Biobank are:

1) To collect and store clinical and genetic information and biological material and their derivatives to allow for future research on SCCOHT and *SMARCA4* gene variants;
2) To connect individuals who are interested in learning more about potential research opportunities with the researchers performing these studies.

The Registry/Biobank will help researchers better understand SCCOHT and may also help researchers develop better screening methods and treatment options for SCCOHT. For rare diseases like SCCOHT, recruitment for studies and clinical trials take a long time. Having a database containing medical information of patients and persons at risk is likely to speed up this process by finding people who may be eligible to participate.

DESCRIPTION OF THE REGISTRY PROCESS

1. Joining the Registry

If you choose to participate in the Registry, you will be asked to provide consent electronically at the end of this form, as well as your contact information (such as your name, address, email address, or phone number). You may choose to provide the contact information of an authorized representative (e.g. spouse, parent, child, sibling etc.) who we may contact in the future in the event that your health status no longer allows you to continue participating in the Registry. In the event that the Registry team does not receive an expected response from you, we will attempt to contact you again every 3

months, up to a maximum of 3 times, then we will stop attempting to contact you again. You are always welcome to contact us at a later date that is more convenient for you.

2. You provide information to the Registry

a) You will be asked to fill out a medical questionnaire for information about your past and present state of health, related but not limited to SCCOHT, genetic testing, demographic information, test results, procedure reports and medical exams. You may not have all the information or be able to answer all of the questions, but we ask you to fill out the questionnaire to the best of your abilities. This may take up to 45 minutes depending on your information. You are encouraged to ask help from your treating physician if needed.

b) The Registry team will send this questionnaire to you by email. You will be able to fill out all questions online and submit them electronically through a secure platform. If you have copies of your medical reports or test results and choose to share these with the Registry, you may do so by uploading PDFs or images of relevant reports directly into the online questionnaire. All information and documents entered into these questionnaires will be transferred securely over an encrypted connection. You may also ask your treating physician or other health care professional to assist with sending medical reports or test results to the Registry.

c) In order to help us complete your questionnaire, you may also choose to authorize the Registry to obtain and review your medical records from your treating institution. If you agree, we will ask for the contact information of your treating physician. In addition to providing your authorization at the end of this consent form, you will likely have to complete a medical record release form with your treating physician/institution in order for us to obtain your records from your treating physician.

3. The Registry staff will review the information you provide, and your information will be stored in our secure databases

The Registry staff will review your responses and may contact you if there are any questions about the information you entered. In order to protect your privacy, your contact information and other identifying information will be coded and stored separately from all other medical information or samples that you provide. Only authorized people who work in the Registry will know the code and be able to identify you if needed.

4. The Registry staff will follow up with you every year to update your information

It is important that the Registry contain accurate and updated information. We encourage you to contact a member of the Registry team to inform us of any major changes regarding your cancer diagnosis/treatments (if applicable), and important test results. If you agree to be contacted by the Registry team in the future, we will email you a reminder to update your Registry information at least once per year. We will send you an update questionnaire containing questions similar to those in the initial survey, but it will be shorter as you will only be asked to provide new information. Additionally, you will be asked to sign an updated medical record release form each year, as needed.

5. The Registry provides researchers relevant information so they can study the data and/or determine who may qualify for their study

In a way that does not reveal your identity, the Registry may share your clinical and genetic data with other researchers (using information that you have entered and/or that comes from medical records

that you have shared with the Registry). Your identifying information (e.g. name, address, phone number, email) will not be shared with anyone outside the Registry. Approved external researchers and clinicians will only be allowed to see de-identified information and may use the de-identified data to search for participants for their studies. If a researcher believes that you qualify for their study, the researcher will inform the Registry's team. If you have agreed to be informed about future research studies that require your direct participation, such as a clinical trial, an authorized member of the Registry team (such as a genetic counselor) will then contact you so that you can decide whether you would like to hear more.

DESCRIPTION OF THE BIOBANKING-RELATED PROCEDURES

You may choose to participate in the Registry without providing samples for the Biobank.
The procedures related to the biobanking will take place at the same place as the rest of the treatments you are undergoing or, if necessary, at a suitable location as determined with you, your physician and the Biobank administrators. The Biobank's team will try to collect biological material during your planned clinical procedures or from excess tissue after such procedures (e.g. request to the pathology department). If this is not possible, you may be asked to provide biological material specifically for the Biobank. You can choose not to provide such material.

You may choose to provide access to the following types of samples for storage in the Biobank:

- **Blood:** In order to avoid multiple blood draws, we can ask that blood samples be taken at the same time as your other study-related or clinical tests. Blood samples collected for the purposes of the biobank will be 10 ml (approximately 2 teaspoons).
- **Tissues:** If your medical care or your participation in a research study involves surgery or a biopsy to remove tissue, the tissue that is not required for diagnosis or hospital archives (excess tissue) will be included in the Biobank. This may include healthy tissue as well as tumor tissue, and may include tissue from past or upcoming procedures.
- **Saliva:** In certain circumstances where obtaining DNA from blood or tissues is difficult or impossible, you may be asked to provide a saliva sample by spitting in a test tube.

What type of research will be done with my information/samples?

Your medical information and samples may be used for any ongoing research about SCCOHT and other related diseases. The material and data collected may be used for genetics-based research. The procedures that the Biobank's team and its collaborators will conduct will be consistent with the standards and technologies available at the time of research. When this Biobank was developed, such procedures included but were not limited to the following methodologies:

- Cytogenetic (including the development of cell lines and tissue-derived models)
    - Using samples that you provide (e.g. blood, tissue), cell lines may be grown and kept in the Biobank. This provides researchers with an unlimited supply of your cells for future studies.
    - Tumour cells may be grown into patient-derived xenografts, organoids or other model systems and stored by the Biobank. These tumour derivatives can be used by researchers to study the tumour's biology, test potential drug treatments, or study drug resistance.
- Biochemical
- Molecular sequencing (DNA or RNA analysis)
    - DNA is a molecule that contains genetic information passed on from one generation to the next. For the purpose of this Biobank, your DNA will be obtained from blood, tissue or saliva.

BENEFITS ASSOCIATED WITH PARTICIPATION IN THE REGISTRY/BIOBANK

You will not directly benefit from your participation in the Registry/Biobank. However, the results of the research projects conducted with your samples and data may lead to better diagnosis and treatments in the future for persons who have the same or a similar condition as you. Additionally, being part of the Registry will allow us to inform you (with your consent) if you are eligible for new clinical trials or research studies that require your direct participation.

Joining the Registry does not mean that you have to join any of the studies you are informed about. After you learn about a specific research or clinical study, you will be able to decide whether you want to participate in the study. However, participation in the Registry does not guarantee you will qualify for, or be enrolled in, all available research or clinical studies.

RISKS ASSOCIATED WITH PARTICIPATION IN THE REGISTRY/BIOBANK

There is minimal risk in taking part in the Registry. The Registry includes questions that can be sensitive, and that you may feel uncomfortable answering. You do not have to share any information you do not want to. A possible risk associated with storing your data is a breach of confidentiality or use of your personal information by an unauthorized third party. To limit this risk, we will take steps to protect your confidentiality described in the Confidentiality section, below.

Risks of sample collection

- Blood draw: Can cause bruising, discomfort, pain, dizziness and rarely, an infection. The amount of blood we collect has been determined to be safe for someone of your age.
- Tissues: There are no additional physical risks to you because samples will only be collected if excess tissue is available after any procedures that are part of your medical care or other research involvement. Extra procedures are rarely done just for the purpose of this Biobank; no procedure to collect tissue will be done without your consent and review of the related risks.
- Saliva Sample: There are no known medical risks in providing a saliva sample**.**

Risks linked to genetic research

Under Canadian law, the Genetic Non-Discrimination Act (GNA) protects individuals from the use of genetic test results in areas outside of medical care and medical research, such as insurance and employment. A similar act exists in the USA (Genetic Information Non-discrimination Act) and the European Union (General Data Protection Regulation). If you reside in a country not covered by such laws and you request that validated genetic results be disclosed to you, there could be a risk that the information returned to you could compromise or diminish your ability to obtain certain services for yourself and your family in your country. However, all genetic data stored on our Registry servers in Canada remain protected by the GNA.

VOLUNTARY PARTICIPATION AND THE RIGHT TO WITHDRAW

Your participation in the Registry/Biobank is voluntary. Therefore, you may refuse to participate. Your decision to not participate, or to withdraw, will have no impact on the quality of care and services to which you are otherwise entitled, or on your relationship with the Registry/Biobank director or the clinical team. Should you withdraw your consent, any material or data that has not been used in research projects will be destroyed. If research has already been conducted and published with your

material or data before you decide to withdraw, it will not be possible to destroy this information.

Any findings that could influence your decision to have your data and biological samples kept in the Registry/Biobank will be shared with you as soon as possible.

CONFIDENTIALITY

Only information required to meet the scientific goals of the Registry/Biobank will be collected about you and added to the Registry/Biobank. All the information collected for the Registry/Biobank will only be used as planned for in its management framework. Beyond those uses, the information collected will remain strictly confidential to the extent provided by law. In the Registry/Biobank, you will only be identified by a code number; your name will not be used. Your samples will also be sent from your treating institution and may contain identifiers at the time of shipment that will be coded once received and processed by the Biobank. The key to the code linking your real identity to your Registry file/Biobank samples will be kept by Dr. William Foulkes, Research Institute of the MUHC.

The Registry/Biobank may share your de-identified samples and/or data with other researchers for research that is not known at this time. You will not be specifically informed if/when your samples and/or data are shared with third parties and if/when research is conducted on them. The Registry/Biobank owner will ensure the confidentiality rules in effect in Quebec and Canada are respected, regardless of the country to which your samples and/or data are then transferred. For example, this means the Registry/Biobank owner will require that third parties do not attempt to re-identify you prior to sharing your samples and/or data with them.

The data may be published or shared during scientific meetings; however, it will not be possible to identify you.

For monitoring, control, safety, security, the Biobank infrastructure, including your samples and data, may be examined by a person mandated by Canadian or international regulatory authorities, such as Health Canada, as well as by representatives of the study sponsor, the institution, or the research ethics board that provides oversight of the biobank (MUHC-REB). All these individuals and organizations adhere to policies on confidentiality.

What if the researchers discover something about me?

During studies, researchers may learn something about you that they didn't expect that may have significant implications for your current or future wellbeing, or that of your family members. For example, the researchers may find out that you have or are at risk for another medical condition. These types of findings are called secondary or incidental findings.

In the event that a significant incidental finding is identified in the course of research with biological material from the Biobank, the decision of whether you and your treating team should be notified by the Biobank will be made according to applicable guidelines. You will be asked to make a decision about these types of findings later on in this consent form.

LOCATION AND LENGTH OF CONSERVATION

Your samples and information will be stored in the Registry/Biobank situated within and under the

auspices of the MUHC Department of Specialized Medicine, Division of Medical Genetics. The samples and data you provide will be stored for as long as the SCCOHT/SMARCA4 Registry and Biobank has a scientific interest for the community and the Director or administrator can ensure its management, starting from the date you provide them.

MARKETING POSSIBILITIES
Research conducted on your banked samples and data will not be used for commercial purposes or for-profit activities.

FUNDING OF THE RESEARCH PROJECT
The SCCOHT/SMARCA4 Registry and Biobank is funded by the Eve Appeal (2020-2022), the Department of Defense Congressionally Directed Rare Cancers Research Program, USA (2021-2024), as well as the Small Cell Ovarian Cancer Foundation and the Research Institute of the McGill University Health Centre. Additional research studies will be linked to the biobank over time to contribute to its continued funding. There is no cost to you to participate.

COMPENSATION
No monetary compensation is associated with your participation in the Registry/Biobank. You will not be reimbursed for the costs you may incur by participating in the biobank (e.g., parking, meals, etc.).

SHOULD YOU SUFFER ANY HARM
Should you suffer harm of any kind following the collection of data and samples for this Registry/Biobank, the Registry is not responsible for any related costs, but you will receive the appropriate care and services required by your state of health, as you would for any other adverse health issue.

By agreeing to participate in the Registry/Biobank, you are not waiving any of your legal rights nor discharging the study doctor, the sponsor or the institution, of their civil and professional responsibilities.

CONTACT INFORMATION
If you have questions or if you have a problem you think may be related to your participation in this Registry/Biobank, or if you would like to withdraw, you may communicate with Dr. William Foulkes, the researcher responsible for this project, by email (william.foulkes@mcgill.ca), or with someone on the study team by phone (514-934-1934 ext 42032) or email (foulkeslab.oncology@mcgill.ca).

For any question concerning your rights as a research participant taking part in this study, or if you have comments, or wish to file a complaint, you may communicate with the Patient Ombudsman of the McGill University Health Centre at the following phone number: 514-934-1934 ext 48306.

OVERVIEW OF ETHICAL ASPECTS OF THE RESEARCH
The Registry/Biobank is hosted at the MUHC and, as such, the MUHC Research Ethics Board reviewed its management framework and is responsible for monitoring it.

Centre universitaire de santé McGill | McGill University Health Centre

INFORMATION AND CONSENT FORM

Registry and Biobank participation

Adult Group

Research Project Title: SCCOHT/SMARCA4 Registry and Biobank

This Registry is be governed by, and construed in accordance with, the laws and regulations of the province of Québec, and the federal laws and regulations of Canada applicable therein. All disputes will be referred to the courts of the Province of Quebec, which will have jurisdiction. By signing this consent form, you irrevocably submit to the jurisdiction of such courts.

Signature of the participant (or legal representative)

I have reviewed the information and consent form. Both the research project and the information and consent form were explained to me. My questions were answered, and I was given sufficient time to make a decision. After reflection, I consent to participate in this research study in accordance with the conditions stated above.

1) I authorize the Registry/Biobank team to have access to my medical records for the purposes of this registry. Yes☐ No☐

   I understand that the Registry/Biobank team will contact my treating physician to obtain relevant medical records and complete my medical information for the Registry. Yes☐

Treating physician: Institution address:

Phone number:

Email:

2) I agree to be contacted by the Registry/Biobank team to update my Registry information annually. Yes☐ No☐

3) I agree to be contacted by the Registry/Biobank team to be informed of any future clinical trials or other studies requiring my direct participation for which I may be eligible: Yes☐ No☐

4) As for the Biobank, I allow access to the following biological samples, if applicable, as explained in this consent form:
   - Blood: Yes☐ No☐
   - Tissue from surgeries or biopsies (*past or upcoming)*: Yes☐ No☐
   - Saliva sample: Yes☐ No☐

5) In the case where there are validated results with possible impact for my health or that of my family member and for which preventive measures or treatment are available, I would like to be informed through a genetic counselor. Yes☐ No☐

6) I agree that my treating physician should be made aware of incidental findings that could be useful to my care or that of my relatives. Yes☐ No☐

Same as above ☐

Treating physician: Institution address:

Phone number:

Email:

If participant is providing consent for themselves:

By checking this box and typing my full name below, I am electronically signing this consent form ☐

Name of participant Date

If consent is being given by a family member or guardian of the participant (the person legally responsible for the care and maintenance of the participant) or on behalf of a deceased individual:

By checking this box and typing my full name below, I am electronically signing this consent form ☐

Name of authorized representative Date

Signature of the person obtaining consent

I have explained the Registry/Biobank and the terms of this information and consent form to the research participant and/or their authorized representative, and I answered any questions they asked.

Name of the person obtaining consent Signature Date