A. PURPOSE OF THE STUDY:
You are being asked to volunteer in a research study. The purpose of this study is to obtain information about how a specific change or alteration in one or more genes increases or decreases a woman's risk for developing breast cancer. The long-term goal is to develop better methods of early breast and ovarian cancer diagnosis, treatment, and prevention. You are being asked to participate in this study because:

1) You have a family history of breast or ovarian cancer which suggests that you may have an increased risk for developing one of these cancers, but you do not have one of the defined mutations (abnormal genes), OR
2) You are 60 years old or older and are known to have a genetic mutation that increases your risk of developing breast or ovarian cancer, although you have not been diagnosed with cancer, OR
3) You have been diagnosed with breast or ovarian cancer and are known to have a defined genetic mutation associated with hereditary breast and ovarian cancer, OR
4) You are 60 years old or older, and you have not been diagnosed with breast or ovarian cancer.

B. SUBJECT PARTICIPATION:
We estimate that the following number of subject will enroll in this study:
At this site: 800 Total at all sites: 1200
SUBJECT PARTICIPATION:
❒Inpatient
❒Outpatient
❒Other [healthy subjects, etc.]
Please specify:
Your participation will involve no additional visits.

C. DESCRIPTION OF RESEARCH:
Procedures: Completion of questionnaire, collection of tissue not required for diagnosis, one-time saliva donation of appox. 4mL.

Information about you and your illness, including your age, family's place of origin, history of relatives affected with cancer, stage of disease, and response to surgery, will be obtained at the time that you enroll. Study staff will gather this information from a questionnaire, review of your medical records and possibly one or more telephone calls to you. A saliva sample will then be taken at an office visit with your oncologist or genetic counselor. Once this information has been obtained, we will remove your name, medical record number, birth date and other identifying information that may be linked to your samples.

The study will involve analysis of genetic markers from your saliva that may increase or decrease a woman's risk for developing breast or ovarian cancer. (These markers are called, "DNA, RNA, and protein.") Your genetic makers and those from other women with breast cancer will be compared to the genetic markers from women unaffected by breast cancer. Because your sample cannot be linked to your name, we will not be able to provide you with results. Nonetheless, once this study has been completed, we may invite you and your family to participate in a follow-up study of the usefulness of genetic testing for women at risk for breast cancer.

D. COSTS/REIMBURSEMENTS:
All costs associated with your being in this study will be paid by the sponsor. You or your insurance company will be charged or held responsible only for the costs of your routine care (the care you would have received if you were not in this study), but not for the tests or consultations specifically related to the study.

If you do not sign this consent form, you will continue to receive care, but not as a part of this study.

This study is being sponsored by private foundations. Portions of Dr. Ostrer's and his research team's salaries are being paid by this grant.

E. POTENTIAL RISKS AND DISCOMFORTS:
The following risks or discomforts to you may occur by being in this research study.
Procedures to guarantee confidentiality are discussed below.
Among the risks that have been associated with genetic testing are learning about susceptibility to other diseases and possible unfair discrimination in employment and insurance. *None of these risks will exist for you, because it will not be possible to link your name to your results*.

F. POTENTIAL BENEFITS:
There is no direct benefit to you expected from your participation in this study. It is hoped the knowledge gained will be of benefit to others in the future.

G. ALTERNATIVES TO PARTICIPATING IN THIS STUDY:
This is not a treatment protocol. You may choose not to participate in this study or to withdraw at any time without any impact to your care.

H. CONFIDENTIALITY:
If you consent to participate in this research, your personal information will be kept confidential because your name, medical record number, and any other identifiers will be removed. Thus, it will not be possible to report results to you, your physician or to a third party. The sponsor of the study and NYUSM staff working under the direction of the IBRA may inspect records related this study, and will learn about your participation from this signed consent form. The medical record is maintained by your treating physician or hospital, as applicable, and will be subject to New York State and federal laws and regulations concerning confidentiality of medical records.

Other persons and organizations, including co-investigators, federal and state regulatory agencies, and the IRB(s) overseeing the research may receive information about your participation during the course of this study. Except when required by law, study information shared with persons and organizations outside of New York University School of Medicine (NYUSM) will not identify you by name, social security number, address, telephone number, or any other direct personal identifier.

When your study information will be disclosed outside of NYUSM as part of the research, the information that can identify you as listed above will be removed and your records will be assigned a unique code number. NYUSM will not disclose the code key, except as required by law.

Confidentiality of Your Medical Records
Your medical records will be kept in accordance with state and federal laws concerning the privacy and confidentiality of medical information. If your participation in this research is for treatment or diagnostic purposes, the facility in which you are treated may ask you to sign a separate informed consent document for specific procedures or treatment, and that informed consent form may be included in the medical record of that facility. The confidentiality of your medical record is also protected by federal privacy regulations, as described below.

Confidentiality of Your Study Information
Your study records include information that identifies you as a participant and that is kept in research files. We will try to keep this information confidential, but we cannot guarantee it. If data from this study are to be published or presented, we will first take out the information that identifies you.

Retention of Your Study Information
The study results will be kept in your research record for at least six years or until after the study is completed, whichever is longer. At that time either the research information not already in your medical record will be destroyed or information identifying you will be removed from such study results at NYU. Any research information in your medical record will be kept indefinitely.

Your HIPAA Authorization
A new federal regulation, the federal medical Privacy Rule, has taken effect as required by the Health Insurance Portability and Accountability Act (HIPAA). Under the Privacy Rule, in most cases we must seek your written permission to use or disclose identifiable health information about you that we use or create [your "protected health information"] in connection with research involving your treatment or medical records. This permission is called an Authorization.

If you sign this form you are giving your Authorization for the uses and sharing of your protected health information described below. You have a right to refuse to sign this form. If you do not sign the form you may not be in the research program, but refusing to sign will not affect your health care (or payment for your health care) outside the study.

This Authorization will not expire unless you withdraw it in writing. You have the right to withdraw your authorization at any time, except to the extent that NYU has already relied upon it or must continue to use your information to complete data analysis or to report data for this study. The procedure for revoking your authorization is described below in Section K.

By signing this form you authorize the use and disclosure of the following information for this research:

- Your medical records
- Your research records
- Results of laboratory tests
- Clinical and research observations made during your participation in the research.

By signing this form you authorize the following persons and organizations to receive your protected health information for purposes related to this research:

- Every health care provider who provides services to you in connection with this study
- Any laboratories and other individuals and organizations that analyze your health information in connection with this study in accordance with the study's protocol
- The following research sponsors and the people and companies they use to oversee, administer, or conduct the research: NIH

- The United States research regulatory agencies and other foreign regulatory agencies
- The members and staff of the hospital's affiliated Institutional Review Board
- The members and staff of the hospital's affiliated Privacy Board
- Principal Investigator: Harry Ostrer, M.D.
- Study Coordinator
- Members of the Research Team
- Members of the NYU/NYUMC Clinical Trials Office/Office of Research and Sponsored Programs
- Data Safety Monitoring Board/Clinical Events Committee

If any of the companies or institutions listed above merges or is sold during the course of this research, your Authorization will cover uses and disclosures of your protected health information to the new company or institution that assumes responsibility for the research.

Please be aware that once your protected health information is disclosed to a person or organization that is not covered by the federal medical Privacy Rule, the information is no longer protected by the Privacy Rule and may be subject to redisclosure by the recipient.

I. COMPENSATION/TREATMENT IN THE EVENT OF INJURY:
All forms of medical (or mental health) diagnosis and treatment – whether routine or experimental – involve some risk of injury. In addition, there may be risks associated with this study that we do not know about. In spite of all precautions, you might develop medical complications from being in this study. If you sustain any injury during the course of the research or experience any side effect to a study drug or procedure, please contact the Principal Investigator, Harry Ostrer, M.D., at the following telephone number 212 263-7596. If such complications arise, the study doctor will assist you in obtaining appropriate medical treatment but this study does not provide financial assistance for medical or other injury-related costs. You do not give up any rights to seek payment for personal injury by signing this form.

J. VOLUNTARY PARTICIPATION AND AUTHORIZATION:
Your decision as to whether or not to take part in this study is completely voluntary (of your free will). If you decide not to take part in this study it will not affect the care you receive and will not result in any loss of benefits to which you are otherwise entitled.
You will be told of any significant new findings developed during the course of the research that may influence your willingness to continue to participate in the research.
Your decision as to whether to give your authorization for the use and disclosure of your protected health information for this study is also completely voluntary; however, if you decline to give your authorization or if you withdraw your authorization you may not participate in the study.

K. WITHDRAWAL FROM THE STUDY AND/OR WITHDRAWAL OF AUTHORIZATION:
If you decide to take part in the study, you may withdraw from participation at any time without penalty or loss of benefits to which you would otherwise be entitled. You may also withdraw your Authorization for us to use or disclose your protected health information for the study.
If you do decide to withdraw your consent, we ask that you contact Dr. Harry Ostrer in writing and let him know that you are withdrawing from the study. His mailing address is NYU School of Medicine, 550 First Avenue, MSB136, New York, NY 10016. If you wish to withdraw your Authorization as well as your consent to be in the study, you must contact Dr. Ostrer in writing. Remember that withdrawing your Authorization only affects uses and sharing of information after your written request has been received, and you may not withdraw your Authorization for uses or disclosures that we have previously made or must continue to make to complete analyses or report data from the research.
The Principal Investigator or another member of the study team will discuss with you any considerations involved in discontinuing your participation in the study. You will be told how to withdraw from the study. The study doctor may also decide to withdraw you from the study for certain reasons. Some possible reasons for withdrawing a subject from the study would be inadequate tissue sample that precludes carrying out the analyses proposed. It will not be possible for you to withdraw your tissue or DNA samples from this study, because it will no longer be possible to identify their being yours.

L. **PERMISSION TO CONTACT YOU ABOUT FUTURE RESEARCH:**
I authorize the principal investigator and his or her coinvestigators to contact me about future research within the NYUSM facility provided that this future research is approved by the original IRB of record and that the principal investigator and coinvestigator are affiliated with the research protocol. If I agree, then someone from Dr. Ostrer's staff might contact me in the future and he or she will tell me about a research study. At that time, I can decide whether or not I am interested in participating in a particular study. I will then have the opportunity to contact the researcher to schedule an appointment to be fully informed about the research project.

Your permission to allow us to contact you about future research would be greatly appreciated, but it is completely voluntary. If you choose not to allow us to contact you, it will not affect your care at any of the NYUSM facilities. Please understand that giving your permission to do this is only for the purpose of helping us identify subjects who may qualify for one of our future research studies. It does not mean that you must join in any study.

M. CONTACT PERSON(S):
For further information about your rights as a research subject, or if you are not satisfied with the manner in which this study is being conducted and would like to discuss your participation with an institutional representative who is not part of this study, please contact the Administrator, Institutional Board of Research Associates, Telephone No. 212-263-4110.

If you have any questions or sustain any injury during the course of the research or experience any adverse reaction to a study drug or procedure, please contact the Principal Investigator Harry Ostrer, M.D. at the following telephone number 212 263-7596.

WHEN THE SUBJECT IS AN ADULT:

Notice Concerning HIV-Related Information: If you are authorizing the release of HIV-related information, you should be aware that the recipient(s) is prohibited from redisclosing any HIV-related information without your authorization unless permitted to do so under federal or state law. You also have a right to request a list of people who may receive or use your HIV-related information without authorization. If you experience discrimination because of the release or disclosure of HIV-related information, you may contact the New York State Division of Human Rights at (212) 480-2493 or the New York City Commission of Human Rights at (212) 306-7450. These agencies are responsible for protecting your rights.

* For subjects who may not be capable of providing informed consent, the signature of a legal representative is required. For a valid HIPAA authorization, the "personal representative" must have authority under state law to make health care decisions for the subject.** When the elements of informed consent are presented orally to the subject or representative, a witness to the oral presentation is required.

INFORMED CONSENT FORM TO PARTICIPATE AND AUTHORIZATION FOR RESEARCH

Genetic Modifiers of Breast and Ovarian Cancer Risk

Principal Investigator:

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