COLUMBIA UNIVERSITY MEDICAL CENTER

CONSENT FORM and HIPAA Authorization

Study Title: Folks-At-Risk for Interstitial Lung Disease (FAR-ILD)

Study number: IRB-AAAR1916
Anticipated number of subjects: 800
Principal Investigator: Christine Kim Garcia MD on behalf of the following investigators: Michaela Anderson MD; Matthew Baldwin MD; Keith Brenner MD; William Bulman MD; Wellington Cardoso, MD, PhD; Sean Fedyna MD; Claire McGroder MD; Nina Patel, MD; Da Zhang MD; Belinda D'Souza MD; Mary Salvatore MD; Anjali Saqi MD; and David Goldstein PhD
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Introduction

The purpose of this form is to give you information to help you decide if you want to take part in a research study. This consent and HIPAA authorization form includes information about:
- Why the study is being done;
- The things that you will be asked to do if you are in the study;
- Any known risks involved;
- Any potential benefit;
- Options, other than taking part in this study, that you have; and
- The way your health information will be used and shared for research purposes.

The principal investigator (the lead researcher for this project, a co-investigator (another researcher involved in this project), or a study coordinator will discuss this study with you. If at any time you have questions about the study, please ask a member of the study team. Take all the time you need to decide whether you want to take part in this research study.

The purpose of this research is described below in the 'What is Involved in This Study?' section of this consent form.

What information is on this form?

We are asking you to take part in a research study. This form explains why we are doing this study and what you will be asked to do if you choose to participate in it. It also describes the way we would like to use information about you and how we would like to use the blood and tissue samples we obtain from you. Please take the time to read this form. We will also talk with you about taking part in this research study. If at any time you have questions about this for or the research study, please ask a member of the study team. Take all the time you need to decide whether you want to take part in the research study. Participation is voluntary; you do not have to participate if you do not want to.

Why is this study being done?

The purpose of this study is to gain a better understanding of the cause of interstitial lung disease (ILD), including the use of genetic testing. You have been asked to participate in the study either because you have been diagnosed with ILD or because you are at risk for developing ILD. We will study the results of the genetic tests being performed to find and possibly confirm associations between ILD and specific genes or genetic variants. We are collecting DNA/RNA samples and clinical information from individuals who have been diagnosed with ILD as well as from individuals at risk for developing ILD. If you agree to participate, your samples and clinical information will be stored at Columbia University Medical Center (CUMC) and will be studied with the goal of identifying causes and contributors to these conditions.

At-Risk Family Members. We are assessing lung health in adults who have another family member with ILD. Participants over the age of 40 years of age will undergo a CAT scan of their chest to assess their lung health. Participants over the age of 35 years of age will undergo breathing tests to assess their lung health.

At-Risk Smokers. We are assessing lung health for adults. Participants will undergo a CAT scan of their chest to assess their lung health. At-risk smokers are eligible to participate if they are 50 year of age and above with a minimum of 30 pack-year smoking history. Participants who have received a diagnosis of chronic obstructive pulmonary disease "COPD: are eligible to participate. However, participants who have been diagnosed with bronchiectasis will not be eligible to participate

At-Risk COVID-19 Survivors. We are assessing lung health of adults who have survived a COVID-19 pneumonia. Participants over the age of 21 years of age will undergo a CAT scan of their chest to assess their lung health. Participants will also undergo breathing tests.

Controls. You might be asked to participate as a control if you are a household contact of someone who is recruited to participate in this study.

What will I be asked to do if I choose to be in this study?

Patients with ILD:
- We will collect medical information and CAT scan images from your medical record.
- We will collect a sample of blood.
- We may collect a nasal swab.
- If you have undergone a biopsy of your lung, we may use some of the lung tissue for research.
- We may ask you to return to see us each year.
- The study will follow-up with you for up to 20 years.
- Patients with ILD who are not being treated at New York Presbyterian/Columbia University Medical Center will be asked to sign a medical release form, the information obtained will be used for research purposes only.

At-Risk Family Members and Survivors of COVID-19 lung disease:
- We will ask you to come to New York Presbyterian/Columbia University Medical Center.
- A doctor will ask you questions about your health and perform a partial physical examination.
- We will collect a sample of blood
- We may collect a nasal swab.

- We will ask you to perform two breathing tests (called pulmonary function tests):
    1. A “spirometry” test to measure the flow of air in and out of your lungs. This will require you to breathe in hard and deep and blow out over a period of time. At least 3 and as many as 6 exhalation efforts will be necessary for this test.
    2. A “diffusing capacity” test to measure how well your lungs exchange oxygen and carbon dioxide. You breathe in and hold your breath for 10 seconds, then rapidly blow it out.
- We will ask you to walk for 6 minutes in a hallway walking test.
- We will ask you to undergo a CAT scan of your chest, which involves radiation.
- We may invite you to undergo a bronchoscopy (described below) to obtain small pieces of your lung for research tests.

At-Risk Participants with Smoking History:
- We will ask you questions regarding your smoking history.
- We will collect a sample of blood.
- We may collect a nasal swab.
- We will ask you to undergo a CAT scan of the chest, which involves radiation. If you have undergone a CAT scan of the chest within the past year, we will ask you to either provide us with a copy of the scan or sign a medical release form so that the research staff can obtain a copy of the scan.
- We may also invite you to undergo pulmonary function testing as well.
- We may invite you to undergo bronchoscopy (described below) to obtain small pieces of your lung for research tests.

Controls:
- We will ask questions about your health and collect information about you. We will specifically ask if you have been diagnosed with a lung ailment.
- We will collect a samples of blood and may collect a nasal swab. These samples may be used for genetic testing and results may be returned to you (see below).
- You will not be asked to undergo bronchoscopy.

Bronchoscopy:
Depending on your test results, we may invite you in to undergo a biopsy of your lung using a procedure called a bronchoscopy, to obtain lung tissue, lung cells, and saline (salt water) washings of the inside of the lungs. A bronchoscopy is a medical procedure performed by a doctor. The bronchoscopy will be performed in a procedure room at New York Presbyterian. If you undergo a bronchoscopy, you will need to have blood drawn again for “routine” blood tests to make sure it is safe to perform a bronchoscopy. If you are a woman, we may ask you for a urine sample to determine whether you are pregnant. You will have an “IV” (intravenous) catheter placed so that medications can be given to you. The study doctor will give you medications that will make you sleepy (“sedation”), but you will not receive general anesthesia as a routine part of this research study. The doctor will put numbing medication (lidocaine) into your nose, throat, and airways in the lungs to limit the amount of cough and discomfort you might experience. The doctor will place a “bronchoscope” through your nose or mouth and then into the airways of your lungs. A bronchoscope is a long, thing, flexible medical device with a camera on the tip. The doctor will be able to see the inside of the airways of your lungs, take pictures, and will also perform the following procedures using the bronchoscope:

1. The doctor will take biopsies of your lung tissue. This means that the doctor will remove small pieces of your lung tissue (the airsacs and airways of your lungs) using cutting devices for research purposes. This tissue will be used for research.
2. The doctor will use a small brush to collect cells from the inside lining of your nose and airways. These cells will be used for research.
3. The doctor will put salt water (saline) into your lung to wash out any cells that may be there. Some of the salt water will be removed by the bronchoscope and used for research.

If there are unexpected findings in your lungs during the bronchoscopy, the study doctor may perform additional procedures (such as more biopsies) to try to find out what these unexpected findings are. You and your doctor will be informed of the results of any additional procedures for unexpected findings.

After the bronchoscopy you will have at least one chest x-ray to make sure the lungs look healthy. A second chest x-ray is sometimes needed. If you undergo bronchoscopy, we will call you one month later to check your health status.

Please write your initials next to the choice you make below:
_____ (initial) yes, I agree to bronchoscopy with biopsy, fluoroscopy and a chest x-ray, which include radiation.
_____ (initial) no, I do not agree to bronchoscopy with biopsy, fluoroscopy and a chest x-ray, which include radiation.

For all Subjects:
Blood samples.
We will ask you to give a blood sample of up to 25mL (about 5 teaspoons) at enrollment and up to 45mL at follow-up visits. The blood will be drawn (taken) using a needle in a vein and sent to be stored in our research laboratory for future genetic testing. If a blood sample cannot be obtained, you will be asked to provide a saliva sample or buccal (cheek swab) sample. Samples collected for this study will be used to extract DNA and RNA. Some of your blood samples will also be stored in Dr. Garcia’s laboratory for future research.

Follow-up evaluations.
We may ask you to return to see us each year, whether you undergo a bronchoscopy or not. The study will follow-up with you for up to 20 years.

Future Use of Data/Specimens
We would like to store the data and biological samples that you agreed to provide as part of this study and use them for future research. They will be stored at CUMC indefinitely either with the researchers on this study or in a central storage facility called a repository. By signing this consent, you agree that your samples can be stored at CUMC indefinitely for future research. Your samples and data will be labeled with a code number that the researchers on this study will be able to link to you. You also agree that your samples can be stored in

de-identified form, which mean that individuals who have access to the repository will not be able to link them to you. By signing this consent, you agree that your data and samples may be used in the future by Dr. Garcia, other CUMC researchers, and researchers at other institutions or commercial entities, that may not yet be identified, for research of ILD or other conditions. If your data and samples are shared with researchers who are not researchers on this study, they will only be given in de-identified form. This means that your name and other identifying information have been permanently removed from your data and samples OR that your data and samples are coded and the researchers who will use them will not have the key to the code. Any future testing or research using your data and samples may lead to the development and use of information, products, tests and treatments having commercial value. You will not receive any compensation that may result from these tests or treatments.

You can change your mind regarding storage and future use of your samples at any time. Please see the Contact section of the consent form for further information.

Long-term storage of your data.
Because our research is funded by various sponsors, we may be required to submit your clinical and genetic data to a data storehouse not maintained by the researchers on this study. We would only submit your data to a storehouse that permits controlled access to the data. This means that researchers who request access to the data must promise that they will protect the data, only share that data as is permitted by the database rules, report any data breeches and not seek to identify any individuals from the data. If your data is submitted to an outside public data storehouse it will be submitted in de-identified form, so that individuals who have access to the database will not be able to link the data to you. Even though we have taken steps to de-identify your data, it may be possible that someone might be able to identify you through your genetic information in combination with data from other public sources. We believe that this is unlikely to happen.

What are the risks of participating in this study?

There may be slight pain or bruising due to the blood draw. We will use only skilled individuals to obtain blood from you.

Pulmonary Function Tests may cause light-headedness, dizziness, and/or syncope.

Even without your name and other identifiers, your genetic information is unique to you. There is a potential risk that someone will identify you from your genetic information or learn something about you by looking at your genetic information; this risk may increase in the future as technologies advance and more researchers study your genetic information. These risks may also affect members of your family. You should discuss your participation in this study with your family and ask the study doctor about any questions or concerns you may have.

The Genetic Information Non-discrimination Act (GINA) is a federal law that prevents insurance companies from using your genetic information to deny health insurance coverage. The law also prevents employers from getting or using genetic information for employment-related decisions. However, the law does not prevent companies that provide life insurance, disability insurance or long-term care insurance from using genetic information.

Current genetic testing is not an exact science, and you should be aware that the genetic testing being done in this study is considered research testing. As with all research, it is possible that although the tests gives us information that we think may be important, we will not know what all of it means. Thus, it is possible that the meaning of the information you are given may change over time as additional research is conducted.

The genetic research may identify genetic changes that may require additional testing to evaluate. This could result in anxiety, uncertainty and additional expenses that may or may not be covered by your insurance. The genetic research may identify serious, untreatable genetic conditions. Such a finding can result in unexpected psychological trauma, both for you and your family. The detection of such a condition could also affect the health or health care needs of your siblings, children or other close relatives. Because we cannot say with certainty how information derived from the genetic research could be used in the future, this study may involve risks that are currently unforeseeable.

Radiation

Radiation is an energy that is all around us. It is in the air, water, food, and ground. This is called natural radiation. Radiation is also made by humans. It is in our buildings and homes. It is also in medical examinations or treatments such as X-rays. The energy used in X-rays and some other types of scans (such as bone density and Positron Emission Tomography, or PET, scans) is known as radiation. Radiation and the risks of receiving radiation are hard to measure. Experts on radiation agree that there is some risk because radiation exposure adds up over our lifetime. You should always carefully think about how much radiation you will receive from medical examinations or treatments. The “effective dose” of radiation is measured in millisievert (mSv) units. On the average, each person in the United States receives about 3.1 mSv/year from natural radiation. People receive about 2.2 mSv/year from flying in airplanes.

This research study includes exposure to radiation. This radiation exposure is not required for your medical care and is for research purposes only. The radiation exposure is necessary to obtain the research information desired. If you have had X-rays in the past or have been exposed in other ways to radiation, or if you think that you might be pregnant, you must tell the investigator before you agree to be in the study.

In this study, you will receive radiation from a chest CT scan. If you are invited to participate in the bronchoscopy sub-study you will also receive radiation from fluoroscopy and a chest x-ray.

The procedure involving radiation from the chest CT scan in this research study will expose you to a very small amount (up to 15 mSv) of radiation. There may be an increase in the chances of your developing cancer many years after this study. The additional risk from this research study is less than 0.3%.

If you are invited to participate in the bronchoscopy sub-study, this research study will expose you to an additional very small amount (1.3 mSv) of radiation from fluoroscopy and a chest X-ray. In this case, the total amount of radiation is 16.3 mSv and the total risk is 0.33%.

At these very low levels, scientists are uncertain as to the actual risk from research and there may be no risk at all.

In addition, the procedures involving radiation in this research study (e.g. fluoroscopy) might increase the possibility of skin injury, hair loss and/or cataracts. Skin injuries seldom occur and are usually limited to a small area of reddening of the skin surface that was irradiated. They rarely result in an ulcer. If hair loss occurs, it is usually temporary, but could be permanent. Cataracts occur rarely and appear many years after the exposure of the eyes to large doses of radiation.

Bronchoscopy and Biopsy

There are risks if you undergo a bronchoscopy and biopsy. Up to 1 out of 50 people who undergo bronchoscopy for a medical reason can expect to have a complication. When complications occur, they are typically minor and are related either to the procedure itself or to medications used during the procedure.

Sedation that you receive during the bronchoscopy can cause low blood pressure, low oxygen levels, high carbon dioxide levels, nausea, vomiting (which can lead to pneumonia).

Topical anesthetics ("numbing medication") can cause seizures, heart rhythm abnormalities, and can affect the ability of your blood to carry oxygen.

Bronchoscopy (along with washings and brushings of the airways) can often cause nasal discomfort, sore throat, chest pain, low-grade fevers, low oxygen levels (requiring the use of oxygen), and pneumonia. Less common complications of bronchoscopy include asthma-like reactions with wheezing, low oxygen levels, fainting, injury or swelling of the voice box in the throat, injury to the airways (bronchi) in the lung, or serious infections. Rarely, the bronchoscopy equipment could malfunction and damage you.

A lung biopsy performed during bronchoscopy increases the risk of the procedure. About 1 out of 30 people will have bleeding and 1 out of 50 will have a collapse of their lung (a "pneumothorax"). Almost all bleeding episodes are mild and resolve spontaneously or with medications. Collapsed lungs are also typically mild and can be treated with oxygen or with a small or large tube placed between the ribs to drain the air. Rarely, one or more life-threatening complications can occur including severe bleeding, severe collapse of the lung, severe abnormal heart rhythms, cardiac arrest (when the heart stops beating), or respiratory failure (when the lungs stop working).

Death is extremely rare, but can occur in up to 1 out of 10,000 bronchoscopies. You should know that we are aware of one healthy person who died after undergoing a bronchoscopy for research purposes. This occurred in 1996 at another hospital not affiliated with our hospital.

Pregnancy/breastfeeding risks

Taking part in this study while you are pregnant could cause harm to your fetus. If you are breastfeeding, taking part in this study could cause harm to your baby. You must not take part in this study if you are pregnant or breastfeeding.

Loss of confidentiality

A risk of taking part in this study is the possibility of a loss of confidentiality. Loss of confidentiality includes having your personal information shared with someone who is not on the study team and was not supposed to see or know about your information. The study team plans to protect your confidentiality. Their plans for keeping your information private are described in the 'confidentiality' section of this consent form.

Incidental Findings

Although the imaging (CAT scan) you will have in this study is being undertaken for research purposes only, it is possible that doctors may notice something that could be important to your health, such as a nodule in your lungs. Although not likely, it is possible that the doctors may notice something that may be very serious and could immediately affect your life. If so, we will contact you to explain what was observed. If you so desire, we will also talk with your private physician. If you do not have a private physician, we will refer you to an appropriate clinic for follow-up. It will be your choice whether to proceed with additional tests and/or treatments to evaluate what we observed, and you or your insurer will be responsible for these costs.

Are there benefits to taking part in this study?

If you agree to take part in the study, and choose to receive results, there may be direct medical benefit to you.

If your CAT scan or biopsy identifies a medical condition, it is possible that your health could be improved by treating that condition.

If a genetic predisposition for a medical condition is found, knowing this information may help determine how to manage your medical care. We hope that in the future, information learned from this study will benefit other people with similar findings.

However, if the sequencing does not find information that would affect your medical care or well being, there may not be any direct benefit to you. The knowledge gained may increase our understanding of genetic testing and results of genetic tests, and help patients in the future.

Confidentiality

To help us protect your privacy, we have obtained a Certificate of Confidentiality from the National Institutes of Health. The researchers can use this Certificate to legally refuse to disclose information that may identify you in any federal, state, or local civil, criminal, administrative, legislative, or other proceedings. For example, if there is a court subpoena, the researchers will use the certificate to resist any demands for information that would identify you, with the following exceptions listed below.

- The Certificate cannot be used to resist a demand for information from personnel of the United States federal or state government agency sponsoring the project and that will be used for auditing or program

evaluation of agency funded projects or for information that must be disclosed in order to meet the requirements of the federal Food and Drug Administration (FDA). You should understand that a Certificate of Confidentiality does not prevent you or a member of your family from voluntarily releasing information about yourself or your involvement in this research. If an insurer, medical care provider, or other person obtains your written consent to receive research information, then the researchers will not use the Certificate to withhold that information.
- The Certificate of Confidentiality will not be used to prevent disclosure to state or local authorities regarding matters such as child abuse and neglect, or harm to self or others.

Any information collected during this study that can identify you by name will be kept confidential. We will do everything we can to keep your data secure, however, complete confidentiality cannot be promise. Despite all of our efforts, unanticipated problems such as a stolen computer may occur, although it is highly unlikely.

If you choose to take part in this study, you are giving us the authorization (i.e. your permission) to use the protected health information and information collected during the research that can identify you. The health information that we may collect and use for this research may include medical information that may be considered sensitive. The research study will not collect HIV test results, history of drug or alcohol abuse, or mental health information.

Information about you may be obtained from any hospital, doctor, and other health care provider involved in your care that is needed for this research purpose.

Any research information that is shared with people outside of Columbia University Medical Center and New York-Presbyterian Hospital will not include your name, address, telephone number or any other direct identified unless disclosure of the information is required by law or you have authorized the disclosure.

Your data, biological samples, questionnaire responses, and health information will be assigned a code number, and separated from your name or any other information that could identify you. The research file that links your name to the code number will be kept in a locked file cabinet and on a password-protected computer. Only the investigator and authorized study staff will have access to the file.

Columbia University Irving Medical Center has recently implemented a new electronic medical record (EMR) system, which will be shared with Weill Cornell Medical Center, and New York-Presbyterian Hospital and its affiliated institutions.

Your participation in this research study will be documented in our new EMR system. Medical records in this system can be viewed by authorized personnel from these institutions. Study monitors and others who provide oversight of the study may also need to access this record.

The following individuals and/or agencies will be able to look at, copy, use, and share your research information.
- The investigator, Columbia University Medical Center, Weill Cornell Medical Center, and New York-Presbyterian Hospital study staff and other medical professionals who may be evaluating the study.
- Staff and authorities from Columbia University, and New York-Presbyterian Hospital, including the Institutional Review Board (IRB)
- The Office of Human Research Protections ('OHRP') and the United States Food and Drug Administration ('FDA')
- National Institute of Health

Your authorization to use and share your health information does not have an expiration (ending) date.

Once your health information has been disclosed to a third party (for example, a pharmaceutical company participating in a study), federal privacy laws may not longer protect it from further disclosure.

You may change your mind and revoke (take back) this consent and authorization at any time and for any reason. To revoke this consent and authorization, you must contact the Principal Investigator,

Dr. Christine Garcia
650 West 168th Street, BB9-903D
New York, NY, 10032

However, if you revoke your consent and authorization, you will not be allowed to continue taking part in the research. Also, even if you revoke this consent and authorization, the researchers and the sponsor (if applicable) may continue to use and disclose the information they have already collected.

What other options are there? You may choose not to take part in this research study.

What if I get hurt while I am on the study?
Taking part in this research study may result in injury or harm to you. In the event of an injury resulting from your participation in this study, you should seek appropriate medical care and inform the study doctor. In the event of an emergency you should go to an emergency room.

If you are injured or harmed as a result of participating in the study and receive medical care through the New York-Presbyterian Hospital (NYPH), a Columbia doctor, or any other health provider, you will be sent a bill for whatever medical care you receive. All or part of your bill may be paid by your health insurance.

Columbia University and the New York-Presbyterian Hospital (NYPH) are not offering to pay you for pain, worry, lost income, the cost of your medical care or non-medical care costs that might occur as a result of your taking parting this study. However, you do not waive any of your legal rights in signing this form.

Will I get compensated?
Patient(s) with ILD:
- You will not receive any payment or compensation for taking part in this study.

Individuals At-Risk for ILD:
If you are eligible for compensation, you will receive your compensation in the form of a gift card.
- You will receive $100 for completing the initial research visit.

- You will receive $50 for completing the annual follow-up research visit.
- You will receive $100 for completing the bronchoscopy portion of the study.

Controls:
-You will receive $25 for completing the visit.

Will I incur costs if I take part in this study?

You will incur no costs to be in this study. You will not be charged for the costs of confirming in a clinical laboratory any genetic findings to be used in research. However, you or your insurance company will be responsible for any additional clinical test, including genetic tests that may be recommended by your physician as a result of information received from the study.

Do I have to be in the study?

Taking part in this study is your choice. You can decide not to take part in or stop being in the study at any time. Your choice will not affect the treatment you receive from doctors and staff at Columbia University Medical Center and New York-Presbyterian Hospital.

Why might researchers want to contact me in the future?

The researchers of this study may want to contact you in the future. They may want to obtain a new sample of blood or saliva or cheek swab, or to clarify information that you have provided, or to obtain updated information about your health, or to provide a preliminary result from this study. By signing this consent form you are giving us permission to contact you in the future for research purposes.

It is possible that in the future, a genetic test could be done on your stored samples that may give results, which could be important to you or a family member's health. Knowing this information could have risks. For example, it may make you anxious. Or, if the results are discovered by insurance companies, it could make it difficult to get certain types of insurance. If Dr. Garcia and the researchers believe that the genetic test results are important and if you agree to be notified about genetic test results, the research study team will contact you. You will have the option to tell us if you want to know about this genetic test result or not.

What are my rights if I take part in this study?

Taking part in this study is your choice. You can decide not to take part or stop being in the study at any time. Your choice will not involve any penalty or loss of benefits to which are entitled and will not affect your access to health care at CUMC or NYPH.

You will need to notify in writing one of the researchers listed in the Contact section of this consent form if you decide to withdraw from the study before it is finished and no longer want to be contacted by the researchers. You will need to specify in your written notice if you want your unused biological samples destroyed and your identifying information removed from all CUMC databases so that your samples and/or data will not be included in any future analyses. However, there are limitations on our ability to exclude your information or remove your biological samples are they have been de-linked from identifying information or deposited in scientific databases, and, if you have given your permission to do so, used or shared with other researchers.

Whom may I call if I have questions?

If you have any questions about this research study, you may contact:

Dr. Christine Kim Garcia by e-mail at ckg2116@cumc.columbia.edu

If you have any questions about your rights as a research participant, or if you have a concern about this study, you may contact the office below.

Human Research Protection Office
Institutional Review Board
Columbia University Medical Center
154 Haven Avenue, 1st Floor
New York, NY 10032
Telephone: (212) 305-5883
E-mail: irboffice@columbia.edu

An Institutional Review Board is a committee organized to protect the rights and welfare of human subjects involved in research.

More information about taking part in a research study can be found on the Columbia University IRB website at: http://www.cumc.columbia.edu/dept/irb

Some Background on Genes

Your entire unique genetic material, made up of DNA, is known as a 'genome.' An 'exome' is the portion of the genome that includes only the DNA that is directly responsible for telling cells how to make the correct parts, or proteins, to function properly.

We would like you to be well informed about genetic research, and for that reason we have, next, a few brief explanations. Please let us know, at any point, if you want or need more information in order to understand. DNA is the material that governs the inheritance of many human traits, such as hair and eye color or the risk of some diseases. DNA is contained in most of the cells that make up the body's tissues. DNA carries the instructions for your body's development and functions. A piece of DNA that determines a specific function of a cell is called a 'gene.' Abnormalities in the information in a gene can lead to disease. Your entire unique genetic material, made up of DNA, is known as a 'genome.' An 'exome' is the portion of the genome that includes only the DNA that is directly responsible for telling cells how to make the correct parts, or proteins, to function properly.

What are Whole Exome Sequencing and Whole Genome Sequencing?

We are requesting your permission to perform genetic testing on your biological samples to identify variants and consider their relationship to interstitial lung diseases. Genetic research is evolving rapidly. We expect that we will perform whole exome sequencing but other genetic tests in addition to, or in the place of, whole exome sequencing (WES) or whole genome sequencing (WGS) may be performed, including new genetic tests that may be developed in the future. WES and WGS are very detailed types of genetic tests. WES searches through the exome for DNA variations that can cause disease. WGS searches through all of the genome, including areas outside of the exome. Because WES and WGS examine a larger portion of the genetic material than traditional tests, they may be able to find causes of disease where other tests did not. WES and WGS may also reveal information about unexpected diseases. Because WES and WGS are more comprehensive than other genetic tests, it is

particularly important that you understand what is involved. The study coordinator will tell you more about WES and WGS. You may also wish to obtain professional genetic counseling prior to signing this consent form. If genetic variants are found, the IGM will make a genetic counselor available to you, your family members and/or your physician in the event that you have additional questions regarding the results of your testing at no cost. However, you or your insurance provider will be responsible for all costs related to your medical care, including clinic visits and medical procedures recommended to you as a consequence of study results.

Will I get a genetic test result?

Genetic information generated by and clinical information recorded as a part of this research study will be analyzed by study investigators in order to identify genetic causes for interstitial lung diseases. Although the goal of the study is to generate as much genetic data as is possible, there is no guarantee that your sample or data will be analyzed. It is highly unlikely that we will discover genetic variants that have such strong effects that they would be considered to be the cause of your disorder therefore you will probably receive no results from this study. However, it is possible that the cause of your ILD might be determined by the research team.

It is possible that a genetic test will give results that the study team believes are linked to some or all features of your disease. It is also possible that a genetic test may predict that you are at increased risk for developing ILD in the future. Knowing this information could have risks. For example, it may make you anxious. Or, if the results are discovered by insurance companies, it could make it difficult to get some kinds of insurance.

Dr. Garcia and the researchers are giving you the option to tell us if you want to be notified about genetic test results. If a genetic test result is found that may impact your medical care and if you want to be notified about genetic test results, you will be contacted that a result has been found that requires further testing. If you are a patient of one of the physicians conducting this study, the research staff will facilitate a referral to a clinic or physician who can order the confirmatory test. Please note that this consent form contains space for you to name a physician you wish to be involved in disclosing results of the confirmatory genetic test you.

You might need to provide an additional blood sample for confirmatory genetic testing. You might be offered genetic counseling as part of this study, or pursue genetic counseling outside of this study. The cost of confirmatory genetic testing is provided by this study.

If you do not have ILD, a positive test might mean that you may be predisposed (i.e., more likely to develop) heart/lung disease. If this is the case, your physician may order additional medical tests. The cost of these additional medical tests are not covered by this study.

If you do have ILD, it is important for you to understand that this study may not identify a cause for your condition because:

- Your ILD is not due to a genetic cause
- A genetic variant exists, but based on current knowledge, it cannot be determined whether it is related to your ILD. In these cases, you will not be informed of any results. Your sample may be used, however, for future tests.

If there is a positive test result, you may want to undergo further independent testing and/or consultation with specialist physicians and/or genetic counselors. Genetic counseling is provided through the study; however independent testing and consultation with a medical geneticist or other medical specialist is not. You should be aware that insurance companies sometimes use information from genetic testing to deny coverage to applicants (see information on GINA above).

Please choose and initial one of the options below to tell us if you want to know about genetic test results now and in the future.

_____ (initial) Yes, please contact me if information is found in studies of my genetic material that would be important to me or my family's personal health. Please note that I wish to include the physician listed below to be involved in disclosing results of confirmatory genetic testing to me. The following physician: _____ (print name) practicing in _____ (city, state) should be involved in disclosing results of confirmatory genetic testing to me.

_____ (initial) No, don't contact me about information found in studies of my genetic material.

Statement of Consent

I voluntarily consent to participate in the study. I have read this consent and HIPAA authorization form which includes information about the nature and the purpose of the study, as well as a description of study procedures.

I have discussed the study with the investigator or study staff, have had the opportunity to ask questions and have received satisfactory answers. The explanation I have been given has mentioned both the possible risks and benefits to participating in the study and the alternatives to participation.

I understand that I am free to not participate in the study or to withdraw at any time. My decision to not participate, or to withdraw from the study will not affect my future care or status with this investigator.

I understand that I will receive a copy of this signed and dated consent form and HIPAA authorization. By signing and dating this consent form, I have not waived any of the legal rights that I would have if I were not a participant in the study.

Study Participant
Print Name _____

Signature _____

Date & Time _____

Person Obtaining Consent
Print Name _____

Signature _____

Date & Time _____