PRINCIPAL INVESTIGATOR: Morgan Similuk, ScM

STUDY TITLE: NIAID Centralized Sequencing Protocol

STUDY SITE: National Institutes of Health (NIH) Clinical Center

Cohort: Standard consent

Consent Version: May 27, 2021

WHO DO YOU CONTACT ABOUT THIS STUDY?

Principal investigator: Morgan Similuk, ScM, 301-435-6691, morgan.similuk@nih.gov

Study coordinator: Thomas Dimaggio, RN, 301-443-8341, thomas.dimaggio@nih.gov

This consent form describes a research study and is designed to help you decide if you would like to be a part of the research study.

You are being asked to take part in a research study at the National Institutes of Health (NIH). Members of the study team will talk with you about the information described in this document. Some people have personal, religious, or ethical beliefs that may limit the kinds of medical or research interventions in which they would want to participate. Take the time needed to ask any questions and discuss this study with NIH staff, and with your family, friends, and personal health care providers. Taking part in research at the NIH is your choice.

If the person being enrolled is a minor, then the term "you" refers to "you and/or your child" throughout this document.

If the person being asked to participate in this research study is not able to give consent to be in this study, you are being asked to give permission for this person as their decision-maker. The term "you" refers to you as the decision-maker and/or the person being asked to participate in this research, throughout this document.

IT IS YOUR CHOICE TO TAKE PART IN THE STUDY

You may choose not to take part in this study for any reason. If you join this study, you may change your mind and stop participating in the study at any time and for any reason. In either case, you will not lose any benefits to which you are otherwise entitled. However, to be seen at the NIH, you must be taking part in a study or are being considered for a study. If you do choose to leave the study, please inform your study team to ensure a safe withdrawal from the research.

WHY IS THIS STUDY BEING DONE?

This study is being done to understand the genetics of health conditions and the immune system, and also participants' perspectives on genetic testing. We will enroll many participants and study many health conditions, including immune conditions and mental illness. We are asking you to

join this study because either you have a condition or trait we study, or you do not and you can help us see how people with and without these conditions are different.

- Genes are made of DNA and are our bodies' instructions. We all have thousands of genes.
- Genetic testing examines DNA "variants," which are differences in genes between two people.
    - We all have lots of variants.
    - Most are harmless. Some cause differences like eye color. However, some cause disease.

What is involved in this study?

Genetic testing:

- We will study your DNA extracted from blood, saliva, or another tissue sample, including previously collected samples we may have stored at the NIH.
- We will look at your DNA in great detail. We are looking for differences in the DNA sequence or structure between you and other people.

Optional survey or interview:

- We may ask you to complete a short survey or interview about your condition and genetics.
- We may follow up with some of you with additional surveys or interviews.
- We will only ask you to complete this survey or interview if you speak English and you are at least 14 years-old.

What are the results of genetic testing?

We will give you results that:

- Are important to your health
- Have been confirmed in a clinical lab
- Suggest that you could be at risk for serious disease that may affect your current or future medical management.

If you are on a study about a disease, then genetic testing may detect information about your/your relative's condition, including results of uncertain importance.

If genetic test results are unrelated to your NIH evaluations, then we will not typically report:

- Normal variants
- Information about progressive, fatal conditions that have no effective treatment
- Carrier status (conditions you don't have but could pass on)

We may not find a genetic cause for your/your relative's condition. Over time, we will re-review the data. If we learn new information important for your health, then we will contact you.

- If we do not find a genetic cause for your condition or trait, then it does not mean your genes do not contribute to your/your relative's condition or trait. We may have missed important variants.
- Because everyone has thousands of variants, the analysis will take months. Further research can take years.

If you are submitting a sample as a relative, your sample may not be fully analyzed. Instead, it may be used for comparison in interpreting your relative's sample. In this case, a full clinical report would not be generated for you, only your relative.

What will happen to stored samples and data?

- We will store your samples and data for future research using a code only the study team can link to you.
- We will keep any information that can be traced back to you as private as possible.
- We will not sell your samples or data. You will not be paid for any products that result from the research.
- If future studies need information (like current health) that we do not already have, then we will contact you.
- In the future, if you change your mind about letting us store your samples, then let us know. We will do our best to destroy your samples but cannot guarantee that we will always be able to do so. However, we cannot destroy any data or remove documents from your NIH medical record.

How will data be shared?

Sharing research helps discovery. We may share samples or data with:

- NIH researchers to help us understand your condition or trait.
- Other researchers, along with a de-identified code and details such as sex, age, and health.
- Large scientific databases, which store de-identified data from thousands of studies.
- We will submit some of your de-identified data to these databases. Your data may be shared broadly for research on any topic. Researchers must apply for access to these databases.
- We may publish results in scientific journals but will not include identifiable information.

What are the risks of this study?

Blood draws: May cause pain, swelling, redness, bruising, lightheadedness, and, rarely, infection. Typically we ask for about half a tablespoon of blood.

Saliva, hair, and nails: May cause discomfort.

Genetic testing: There are psychological risks.

- You may feel upset if we do not find anything.
- You may feel upset if you learn about genetic problems, health risks, or risks to relatives.
- Your family may be upset, too. They may have different views on genetics and what to do.
- While unlikely, we may learn that you are adopted or have a different parent than you think. We do not tell people this unless it means there are risks to their health.

Surveys and questionnaires: If you complete a survey or questionnaire, some of the questions may be upsetting or make you feel uncomfortable. You can skip any of the questions you do not want to answer, and you can stop at any time.

Protections against misuse of genetic information

This study involves genetic testing on samples. Some genetic information can help predict future health problems of you and your family and this information might be of interest to your employers or insurers. The Genetic Information Nondiscrimination Act (GINA) is a federal law that prohibits plans and health insurers from requesting genetic information or using genetic information. It also prohibits employment discrimination based on your health information. However, GINA does not address discrimination by companies that sell life insurance, disability insurance, or long-term care insurance. GINA also does not protect you against discrimination based on an already-diagnosed condition or disease that has a genetic component.

What are the benefits to taking part in this study?

- You may not benefit from this study.
- You may learn important genetic information.
- This study will help scientists better understand the genetics of your/your relative's condition/trait.

What are my other options?

- You do not have to join this study.
- You could have genetic testing through another research study or a commercial lab.
- You could also see other specialists and get their opinions about your condition/trait.

What if I change my mind?

- You may quit this study at any time without it affecting participation in other NIH research.
- If you withdraw from the study, we will continue to store and use your samples and data.
- Additionally, you may be removed from the study without your consent if your study doctor feels it is in your best medical interest, if the scientific goals of the study change, or if the study is cancelled.

What if there are new findings?

Any new findings, including those that may affect your willingness to continue, will be discussed with you.

Carefully consider these issues. Specialized healthcare professionals called genetic counselors, whose job is to help patients understand their genes and health, are available to discuss any other concerns in greater detail.

PAYMENT

Will you receive any type of payment for taking part in this study?

You will not receive payment for taking part in this study.

REIMBURSEMENT

Will you receive reimbursement or direct payment by NIH as part of your participation?

This study does not offer reimbursement for, or payment of, travel, hotel, or meals.

COSTS

Will taking part in this research study cost you anything?

NIH does not bill health insurance companies or participants for any research or related clinical care that you receive at the NIH Clinical Center. We do not pay for your medical care outside the NIH.

CLINICAL TRIAL REGISTRATION AND RESULTS REPORTING

A description of this clinical trial will be available on http://www.ClinicalTrials.gov, as required by U.S. Law. This Web site will not include information that can identify you. At most, the Web site will include a summary of the results. You can search this Web site at any time.

CONFIDENTIALITY PROTECTIONS PROVIDED IN THIS STUDY

Will your medical information be kept private?

We will do our best to make sure that the personal information in your medical record will be kept private. However, we cannot guarantee total privacy. Organizations that may look at and/or copy your medical records for research, quality assurance, and data analysis include:

- The NIH and other government agencies, like the Food and Drug Administration (FDA), which are involved in keeping research safe for people.
- National Institutes of Health Intramural Institutional Review Board

When results of an NIH research study are reported in medical journals or at scientific meetings, the people who take part are not named and identified. In most cases, the NIH will not release any information about your research involvement without your written permission. However, if you sign a release of information form, for example, for an insurance company, the NIH will give the insurance company information from your medical record. This information might affect (either favorably or unfavorably) the willingness of the insurance company to sell you insurance.

All interviews will be recorded and professionally transcribed. Personally identifiable information such as your name will be removed from each interview recording before it is sent to an outside transcription service. Audio recordings of interviews will be permanently deleted once they are transcribed.

If we share your specimens or data with other researchers, in most circumstances we will remove your identifiers before sharing your specimens or data. You should be aware that there is a slight possibility that someone could figure out the information is about you.

Further, the information collected for this study is protected by NIH under a Certificate of Confidentiality and the Privacy Act.

Certificate of Confidentiality

To help us protect your privacy, the NIH Intramural Program has received a Certificate of Confidentiality (Certificate). With this certificate, researchers may not release or use data or information about you except in certain circumstances.

NIH researchers must not share information that may identify you in any federal, state, or local civil, criminal, administrative, legislative, or other proceedings, for example, if requested by a court.

The Certificate does not protect your information when it:

1. is disclosed to people connected with the research, for example, information may be used for auditing or program evaluation internally by the NIH; or
2. is required to be disclosed by Federal, State, or local laws, for example, when information must be disclosed to meet the legal requirements of the federal Food and Drug Administration (FDA);
3. is for other research;
4. is disclosed with your consent.

The Certificate does not prevent you from voluntarily releasing information about yourself or your involvement in this research.

The Certificate will not be used to prevent disclosure to state or local authorities of harm to self or others including, for example, child abuse and neglect, and by signing below you consent to those disclosures. Other permissions for release may be made by signing NIH forms, such as the Notice and Acknowledgement of Information Practices consent.

Privacy Act

The Federal Privacy Act generally protects the confidentiality of your NIH medical records we collect under the authority of the Public Health Service Act. In some cases, the Privacy Act protections differ from the Certificate of Confidentiality. For example, sometimes the Privacy Act allows release of information from your medical record without your permission, for example, if it is requested by Congress. Information may also be released for certain research purposes with due consideration and protection, to those engaged by the agency for research purposes, to certain federal and state agencies, for HIV partner notification, for infectious disease or abuse or neglect reporting, to tumor registries, for quality assessment and medical audits, or when the NIH is involved in a lawsuit. However, NIH will only release information from your medical record if it is permitted by both the Certificate of Confidentiality and the Privacy Act.

POLICY REGARDING RESEARCH-RELATED INJURIES

The NIH Clinical Center will provide short-term medical care for any injury resulting from your participation in research here. In general, no long-term medical care or financial compensation for research-related injuries will be provided by the NIH, the NIH Clinical Center, or the Federal Government. However, you have the right to pursue legal remedy if you believe that your injury justifies such action.

PROBLEMS OR QUESTIONS

If you have any problems or questions about this study, or about your rights as a research participant, or about any research-related injury, contact the Principal Investigator, Morgan Similuk, at 301-435-6691 or morgan.similuk@nih.gov. You may also contact the study coordinator, Thomas Dimaggio, at 301-443-8341 or thomas.dimaggio@nih.gov. You may also call the NIH Clinical Center Patient Representative at 301-496-2626, or the NIH Office of IRB Operations at 301-402-3713, if you have a research-related complaint or concern.

CONSENT DOCUMENT

Please keep a copy of this document in case you want to read it again.

Adult Research Participant: I have read the explanation about this study and have been given the opportunity to discuss it and to ask questions. I consent to participate in this study.

Signature of Research Participant	Print Name of Research Participant	Date

Legally Authorized Representative (LAR) for an Adult Unable to Consent: I have read the explanation about this study and have been given the opportunity to discuss it and to ask questions. I am legally authorized to make research decisions on behalf of the adult participant unable to consent and have the authority to provide consent to this study. As applicable, the information in the above consent was described to the adult participant unable to consent who agrees to participate in the study.

Signature of LAR	Print Name of LAR	Date

Parent/Guardian of a Minor Participant: I have read the explanation about this study and have been given the opportunity to discuss it and to ask questions. I give permission for my child to take part in this study.

Signature of Parent/Guardian	Print Name of Parent/Guardian	Date
Signature of Parent/Guardian	Print Name of Parent/Guardian	Date

Investigator:

Signature of Investigator	Print Name of Investigator	Date

Witness should sign below if either:

1. **A short form consent process has been used to enroll a non-English speaking subject or**
2. **An oral presentation of the full consent has been used to enroll a blind or illiterate subject**

Signature of Witness	Print Name of Witness	Date

NIH ADMINISTRATIVE SECTION TO BE COMPLETED REGARDING THE USE OF AN INTERPRETER:

____ An interpreter, or other individual, who speaks English and the participant's preferred language facilitated the administration of informed consent and served as a witness. The investigator obtaining consent may not also serve as the witness.

____ An interpreter, or other individual, who speaks English and the participant's preferred language facilitated the administration of informed consent but did not serve as a witness. The name or ID code of the person providing interpretive support is: _____.