Research Study Assent: Ages 13-17

IRB# NCR234775

Principal Investigator: Henry Kaminski, MD

Study Title: Genome Wide Association Study of Patients with Muscle-Specific Kinase Myasthenia Gravis

You are invited to participate in a research study under the direction of Dr. Henry Kaminski of the Department of Neurology, George Washington University (GWU), and paid for by National Institutes of Health. Taking part in this research is entirely voluntary.

This study is part of the Rare Diseases Clinical Research Network (RDCRN), an initiative of the National Institutes of Health (NIH) to advance medical research on rare diseases. A long-term goal of the network is to improve diagnosis and treatment of rare disease conditions.

The purpose of this study is to understand and identify differences in genes associated with MuSK myasthenia gravis. We plan to collect saliva samples from 1,000 subjects with MuSK myasthenia to do genetic testing in collaboration with the National Institutes of Health. The expected duration for this study is a one-time submission of saliva sample.

What will happen during this study?

If you agree to be in this study, you will need to:

- contact the study team to learn more about the study
- provide confirmation of your MuSK myasthenia gravis diagnosis.
- undergo informed consent process to sign consent form prior to sample collection
- provide saliva sample within 5 days of receipt of sample package in person or through mail to be returned back to George Washington University.

- **Steps to collect saliva:**

Step 1: Remove the collection funnel and tube from the pack. Next, close the collection funnel on to the tube lightly.
Step 2: Spit saliva into funnel until saliva liquid (not foam) reaches fill line.
Step 3: Untwist the collection funnel and replace with the tube cap tightly. Throw away the funnel.
Step 4: Shake tube a few times to mix the saliva with the solution.

Note: Saliva collection may seem simple, but does take time and effort to fill the small container to the fill line. The liquid part of the saliva needs to be at the fill line and bubbles do not count. This process often takes several minutes to produce enough saliva and people with a dry mouth may take longer.

What are the study risks?

There are no physical risks associated with this study. However, there is a potential risk of loss of confidentiality. Research staff will make every effort to keep your information confidential. Information from this research project will be maintained in a secure GW MFA server at the George Washington University under the supervision of Dr. Henry Kaminski. If results of this research study are reported in journals or at scientific meetings, the people who participated in this study will not be named or identified.

What are the study benefits?

There is no direct benefit for providing one-time saliva sample for this research project. Research conducted using your sample will help to better understand causes for MuSK myasthenia and ultimately provide insight for targeted treatment.

How will my privacy and health information be protected?

Limited demographic data (name, DOB, sex at birth, race, ethnicity) and clinical data (medical history and family history), will be collected by the George Washington University research team under supervision of Henry Kaminski, MD. This limited data will be stored in a primary research database at the RDCRN Data Management and Coordinating Center (DMCC) at the Cincinnati Children's Hospital designated by the National Institutes of Health.

You will be assigned a subject identifier or ID as part of the study and a code number called a Global Unique Identifier (GUID) using an NIH GUID system. The GUID is a unique code made up of letters and numbers that allows researchers to share data from other studies in which you have participated without letting others know who you are. A GUID does not contain personal information, and you cannot be identified using only the GUID. We will ask you for your full date of birth (day, month, year), first name at birth, last name at birth, middle name at birth (if applicable), city/municipality of birth, country of birth to create your GUID.

We would like to make your data, without direct personal identifiers, available for other research studies that may be done in the future. The data/samples without direct identifiers collected from this study will be shared and stored indefinitely at the NIH lab of Dr Traynor. Any personal identifiable information will not appear in the federal data repository or this study clinical research database with exception of e-consent option maintained by the RDCRN DMCC which includes your name and contact information. This information will be stored to document electronic consenting process and to contact study team if you have any questions. A code link with your name and other identifying information will be kept in a secure, password protected file under GW MFA servers maintained by GW research team under supervision of Dr. Henry Kaminski.

Our goal is to make more research possible to learn about health and disease. Researchers who want access to your data will have to request from the NIH. The NIH and the data management center uses several layers of protection for the clinical data stored there

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 except cannot be used to resist a demand for information from personnel of the United States Government that is used for auditing or evaluation of Federally funded projects or for information that must be disclosed in order to meet the requirements of the federal Food and Drug Administration (FDA). The Certificate of Confidentiality will not be used to prevent disclosure to state or local authorities for reasons such as child abuse and neglect, or harm to self or others.

The Health Insurance Portability and Accountability Act (HIPAA) requires that researchers and health care providers protect the privacy of information that identifies you and relates to your past, present and future physical and mental health or conditions, or the provision of health care.

Protected health information that may be used and released (disclosed) in this study includes information such as:

• This consent form
• Demographic information (like your name, date of birth, etc.)
• Information about your medical history from your medical records and your doctor's office
• Laboratory results obtained on specimens collected from you (your saliva sample);
• Interviews with you conducted by members of the Research Team
• Other data created or collected during this study

The researcher and the other members of the research team may obtain your individual health information from hospitals, clinics, health care providers, and health plans that provide health care to you for the purpose of the study.

By signing this form, you allow the use, sharing, copying, and release of your protected health information in connection with this study by:

- The members of the research team;
- Other healthcare providers such as labs which are part of the study;
- Institutional officials who are responsible for compliance;

If you decide to take part in this study, your consent for this study will not expire unless you cancel (revoke) it. The information collected during your participation will be kept indefinitely. You can always cancel this Authorization by writing to the study doctor. If you cancel Authorization, you will also be removed from the study. However, standard medical care and any other benefits to which you are otherwise entitled will not be affected. Canceling your Authorization only affects the uses and sharing of your information *after* the study doctor gets your written request. Information gathered before then may need to be used and given to others. Once your health information has been disclosed to others outside of the medical practice, the information may no longer be covered by the federal regulation that protects privacy of health information. You can also refuse to sign this consent/Authorization to not be part of the study. You can also tell us you want to leave the study at any time without canceling the Authorization. Finally, you should understand that the study doctor is not prevented from taking steps, including reporting to authorities, to prevent serious harm to yourself or others.

What else should I know about the study?

- Someone will explain this research study to you. You may ask all the questions you want before you decide whether to participate or not.
- Participation is voluntary; whether or not you take part is up to you.
- You can agree to take part and later change your mind.
- Your decision to not take part or to stop your participation will not be held against you.
- Your decision will not affect the medical care you receive. If you decide not to take part, you can still continue to receive medical care.
- You may take this document home to read or to discuss with your family members or doctor before deciding to take part in this research study.

May we contact you about future studies that may be of interest to you?

____ Yes ____ No

Electronic Consent (eConsent) option

If you choose to use the electronic consent option instead of paper option, you will be directed to a website and database administered by the Data Management and Coordinating Center (DMCC) at the Cincinnati Children's Hospital. You will be asked to enter your name and contact information into the electronic website. This information will be stored to document the electronic consenting process and to contact you if you indicate you have any questions.

Sample for Genetic Testing

Dr. Traynor at the National Institute of Aging at the NIH will perform genetic analysis or genetic testing on your sample. Genetic research is a research that studies genes, including gene characteristics that are passed down by parents to children. Genetic research may include looking at information, such as personal appearance and biochemistry, gene sequences, genetic landmarks, individual and family medical histories, reactions to medications and responses to treatment. Genetic research raises certain questions about informing you of any results. Possible risks of knowing results include: anxiety; other psychological distress; and the possibility of insurance and job discrimination. A possible risk of not knowing includes being unaware of the need for treatment. These risks can change depending on the results of the research and whether there is a treatment or cure for a particular disease.

Sometimes patients have been required to furnish information from genetic testing for health insurance, life insurance, and/or a job. A Federal law, the Genetic Information Nondiscrimination Act of 2008 (GINA), generally makes it illegal for health insurance

companies, group health plans, and employers with 15 or more employees to discriminate against you based on your genetic information.

Who should I ask if I have any questions?

If you have any questions, concerns or complaints about the study, contact the study team, listed below:

	Study Doctor	Study Coordinator
Name:	Henry Kaminski, MD	Helen Girma
Address:	2150 Pennsylvania Ave NW 7th Floor Washington DC 20037	2150 Pennsylvania Ave NW 7th Floor Washington DC 20037
Telephone number:	202-741-2710	202-677-6205
Email address:	hkaminski@mfa.gwu.edu	hgirma@mfa.gwu.edu

If you have any questions, concerns or complaints about your rights as a research participant and the research, The Office of Human Research of George Washington University, at telephone number (202) 994-2715, can provide further information.

Do I have to be in the study?

No, you do not have to be in the study. Even if you say yes now, you can change your mind later. It is up to you.

Signatures

Before deciding if you want to be in the study, ask any questions you have. You can also ask questions during the time you are in the study.

If you sign your name below, it means that you agree to take part in this research study.

_____	_____
Your Name (Printed)	Age
_____	_____
Your Signature	Date
_____	_____
Signature of Person Obtaining Consent	Date

Signature Block for Adult

By signing below, you agree that the above information has been explained to you and you have had the opportunity to ask questions. You understand that you may ask questions about any aspect of this research for your child during the course of the study and in the future. Your signature below documents your permission for your child to take part in this research.

_____

Printed name of child

_____ _____

Signature of Parent(s) or Legal Guardian Date

_____ _____

Signature of person obtaining consent Date