Version Date: 5/15/17

Permission to Take Part in a Human Research Study

PROTOCOL TITLE: Genomic Medicine at Northwestern: Discovery and Implementation in the eMERGE Network

PRINCIPAL INVESTIGATOR: Maureen Smith, MS

SUPPORTED BY: Northwestern University, Northwestern Medicine, National Human Genome Research Institute (NHGRI)

Why am I being asked to take part in this research study?

You are being asked to be in this study because you are a patient at Northwestern Medicine. This document has important information about the reason for the study, what you will do if you choose to be in this research study, and the way we would like to use information about you and your health.

What should I know about a research study?

- Someone will explain this research study to you.
- Whether or not you take part is up to you.
- You can choose not to take part.
- You can agree to take part and later change your mind.
- Your decision will not be held against you.
- You can ask all the questions you want before you decide.

Who can I talk to?

If you have questions, concerns, or complaints, or think the research has hurt you, talk to the research team:

Sharon Aufox and Christie Hoell are the study managers for this study and can be reached at (312) 503-6200, Monday-Friday 8:30am-5:00pm.

This research has been reviewed and approved by an Institutional Review Board ("IRB"). You may talk to them at (312) 503-9338 or irb@northwestern.edu if:

- Your questions, concerns, or complaints are not being answered by the research team.
- You cannot reach the research team.
- You want to talk to someone besides the research team.
- You have questions about your rights as a research participant.
- You want to get information or provide input about this research.

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Why is this research being done?

New technologies have helped us begin to understand the genetics of many health conditions. This knowledge has opened up the possibility of testing people before they start showing symptoms of specific conditions. Recently, recommendations have stated that anyone having one of these genetic tests must have certain results be returned to them, even if those results are not related to the reason the person had the testing in the first place or is unrelated to a condition the patient has. These results can be related to different types of conditions such as certain types of heart disease and cancers. We are interested how you will understand and use the information both in your personal life and in making health care decisions.

Research into genetics and its influence on health conditions involve the study of differences found in our genes. Genes are the biological instructions that tell our bodies how to grow and function. Genes are made up of building blocks called DNA. The type of genetic testing we will be doing is called sequencing. Sequencing is the process of figuring out how the DNA is arranged in a gene. Learning about the DNA sequence can give us clues about how our bodies grow and function.

How long will the research last?

We expect that this study will last approximately 3 years.

How many people will be studied?

We expect about 3000 patients will be enrolled at Northwestern.

What happens if I say "Yes, I want to be in this research study"?

All participants will have their blood drawn and undergo genetic testing. This testing will look for genetic changes in approximately 130 genes that may influence the development of certain health conditions. However, this testing will not detect all the changes in these genes. Information we give you about your genetic test results will be based on what is known to us at the time of the study. Some of the conditions being tested for have symptoms where treatment is available; other conditions being tested may not.

The genetic results from this testing will be stored in your electronic medical record at Northwestern and your Northwestern healthcare provider(s) will have access to them. If your physicians choose, your results may also be available to you through your MyChart account.

You will receive results related to genes that professional organizations have recommended be returned to patients. You will also be tested for some additional genes that we are interested in studying. Some of those genes are still being studied for how they cause disease.

If the testing does find a change that could potentially impact your health and/or healthcare, you will be notified by a physician or genetic counselor associated with the study either by phone, email, mail, or through your MyChart account about your results. Only approximately 5% of participants in this study are expected to have a genetic difference identified that impacts their health. For those genes where the link between a genetic difference and disease are still being studied, those results will not be placed in your medical records. We expect that it could take up to one year for you to receive your results.

If you consent to participate, you **will** be agreeing to:

- Give about 1 tablespoon (10mls) of blood
- Complete a 20 minute survey that will ask you questions about your expectations about the results you will receive, potential impact of the results, and who you might share your results with
- Provide permission to place the results of your genetic testing in your medical record at Northwestern
- Allow ongoing access to your medical record at Northwestern for the length of the study

If you consent to participate, you **might** be asked to:

- Be interviewed and/or sent a survey after receiving your results about how you used or plan to use the information, how the results may have impacted your health decisions, and any value you placed on these results
- Answer questions about the health of family members

Not all participants will be contacted to be interviewed or to provide additional information.

You will also be given the **option** to:

- Sign up for MyChart. We ask that you consider activating your MyChart account, which is a secure, online access way to access portions of your medical record, communicate with your healthcare providers at Northwestern, and access trusted health information resources
- Participate in in Northwestern's genetic biobank (called the NUgene Project) at the end of this consent form. A genetic biobank is a collection of genetic samples and health information that can be shared with researchers to study the role genes play in human disease.

What happens if I do not want to be in this research?

You can leave the research study at any time and it will not be held against you.

What happens if I say "Yes", but change my mind later?

You can leave the research study at any time and it will not be held against you. If you choose to leave the study prior to the genetic testing, your sample will be destroyed. If you leave the study prior to your results being placed into your medical record, they will not be placed into your medical record. However, once your results are in your medical record, the study cannot remove

them. If you choose to leave the study after results are in your medical record, you will have to follow Northwestern Medicine's procedures for changing information in the medical record.

Is there any way being in this study could be bad for me?

The risks from the study procedures include:

- The risks of taking blood include pain, a bruise at the point where the blood is taken, redness and swelling of the vein and infection, and a rare risk of fainting.

- Disease testing and genetic research may produce results about a current medical condition. Genetic testing may also find that you inherited a genetic change(s) that puts you at risk to develop a condition at some future date. Knowing genetic results has risks. The results may cause you anxiety and other psychological distress, or could also affect your future relationships with family members.

- These results will become part of your medical record. If released, the information could lead to life insurance discrimination, job, or social discrimination. However, there are laws that exist that can protect you from certain types of discrimination. The federal Genetics Information Nondiscrimination Act (GINA) of 2008 offers protection from discrimination based on genetic information by employers and health insurance companies.

- As part of understanding your test results, other family members may be asked to participate in this study. If other family members participate, there is the risk that the results may show that some of your family members are not genetic relatives. There is also a risk that other family members may learn private genetic information about you.

- Not knowing results also has risks. It could mean that you will not have enough information regarding the need for treatment or to estimate the chance to develop a condition.

Will being in this study help me or others in any way?

- It is possible that you may not have any direct benefit from being in this research study.
- Information from this study may help you and your doctors make decisions about how you can manage your health care.
- Taking part in this study may help scientists to better understand how physicians and patients use this information.

What other procedures or courses of treatment might be available to me?

You do not have to take part in this research study. Clinical genetic tests are available for many of the genes being tested for. You may choose to pursue testing on your own. If you choose to not participate in this study, your healthcare providers will make choices about your health care based on current standards of care.

Will it cost me anything to participate in this research study*?*

- No, you will not be charged for any costs related to the study. This includes the blood draw, genetic testing, and genetic counseling.
- The cost of your regular medical care will be billed to you or to your insurance company in the usual way. Any follow-up procedures related to your genetic results that your doctor recommends will be considered part of your regular medical care, and will not be paid for by the research study.

Will I receive payment for participation in this study?

No, you will not be paid for your participation in this project. However, you have the option of being contacted to be interviewed after receiving your results. If you are chosen by the study to be contacted and you agree to be interviewed, you will receive $50 for your time. This payment will either be delivered in person or by mail following the completion of the interview.

What happens to the information collected for the research study?

We respect your privacy. We will keep your personal information confidential. All the information we collect will be kept in secured password-protected servers accessible only by authorized study personnel.

Efforts will be made to limit the use and disclosure of your personal information, including research study and medical records, to people who have a need to review this information. We cannot promise complete secrecy. Organizations that may inspect and copy your information include the IRB and other representatives of this institution.

HIPAA Authorization

We are committed to respect your privacy and to keep your personal information confidential. When choosing to take part in this study, you are giving us the permission to use your personal health information that includes health information in your medical records and information that can identify you. For example, personal health information may include your name, address, phone number or social security number. Your health information we may collect and use for this research includes:

- All information in a medical record
- Results of physical examinations
- Medical history
- Lab tests, or certain health information indicating or relating to a particular condition as well diaries and questionnaires
- Administrative information, such as diagnostic and procedure codes
- Genetic health information related to the test results we place in your medical records

The following groups of people may give the researchers information about you:

- All current and previous health care providers, including but not limited to the Rehabilitation Institute of Chicago (RIC), Northwestern Medical Group (NMG), Northwestern Memorial Hospital (NMH), and Northwestern Lake Forest Hospital.

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Once we have the health information listed above, we may share some of this information with the following people. Please note that any research information shared with people outside of Northwestern University and its clinical partners (or affiliates), the Northwestern University Institutional Review Board Office and Office for Research Integrity, the US Office of Research Integrity, the US Office for Human Research Protections will not contain your name, address, telephone or social security number or any other direct personal identifier unless disclosure of the direct identifier is required by law.

- Authorized members of the Northwestern workforce, who may need to see your information, such as administrative staff members from the Office for Research, Office for Research Integrity and members of the Institutional Review Board (a committee which is responsible for the ethical oversight of the study),

- Clinical affiliates, including but not limited the Rehabilitation Institute of Chicago (RIC), Northwestern Medical Group (NMG), Northwestern Memorial Hospital (NMH), and the Ann & Robert H. Lurie Children's Hospital of Chicago (Lurie Children's).

- Clinical affiliates, including but not limited to Northwestern Medical Group (NMG), Northwestern Memorial Hospital (NMH), and Northwestern Lake Forest Hospital (NLFH) for purposes including, but not limited to, the affiliate's provision of care to you and/or the affiliate's scheduling of appointments and/or billing activities.

- Other University research centers and University contractors who are also working on the study,

- Study monitors and auditors who make sure that the study is being done properly,

- Government agencies and public health authorities, such as the Food and Drug Administration (FDA), the Department of Health and Human Services (DHHS), and the National Institutes of Health (NIH).

- Registries or other research-related databases such as the database of Genotypes and Phenotypes (dbGaP). dbGap is a national genetic database maintained by the National Institutes of Health.

Those persons who get your health information may not be required by Federal privacy laws (such as the Privacy Rule) to protect it. Some of those persons may be able to share your information with others without your separate permission.

The results of this study may also be used for teaching, publications, or presentations at scientific meetings.

Although you may revoke consent to participation in this research at any time and in any format, you must revoke authorization for use or disclosure of your health information in writing. To revoke your authorization, write to:

PI's Name: Maureen Smith

Institution: Northwestern University

Department: Center for Genetic Medicine

Address: 645 N. Michigan Ave. Suite 630, Chicago, IL 60611

You do not have to authorize the use or disclosure of your health information; however, you will not be allowed to take part in this research study. If you do not authorize the use or disclosure of your health information, it will not affect your treatment by health care providers, or the payment or enrollment in any health plans, or affect your eligibility for benefits.

A copy of this signed consent document, information about this study, and the results of any testing done may be included in your medical records and be seen by your insurance company. You will also be provided a copy of this consent form.

Information about the NUgene Project Option

The NUgene Project is a genetic biobank that collects genetic samples and health information that can be shared with researchers. The NUgene Project aims to help researchers better understand health problems and/or the roles genes play in health. It also can help with the development of new treatments or screening tests as we improve our understanding of what causes many health problems. We cannot predict all of the diseases that might be studied using samples from the NUgene Project, but they tend to be diseases that we develop as we get older, such as cancer, heart disease, and diabetes.

If you agree to participate in the NUgene Project, the following will be asked to:

- Give one tablespoon (10ml) of blood. This blood can be obtained when you have you blood drawn for the genetic testing performed for the main study.
- Fill out a brief questionnaire, that asks questions about your and your family's health
- Allow the NUgene Project to have ongoing access to and review your electronic medical records at Northwestern Medicine or its affiliates for as long as you remain in the study.

Unlike the main study, the NUgene Project does not return results from studies that use its samples and samples will not be destroyed unless you withdraw from the project.

The NUgene Project minimizes the risks to privacy and confidentiality by taking the following precautions:

- **The genetic information, research results, and the medical information used in these studies are labeled with a unique number and NOT with any personal identification, so there is little risk that the results can be linked to you.**

- **No information about your participation in the NUgene Project will be placed in your medical records.**

- **The genetic information, questionnaire information, and health information are kept in a secure database or in our collaborators' secure database, separate from any identifying information (such as your name, date of birth, and social security number).**

- **The Principal Investigator of the NUgene Project has obtained a Certificate of Confidentiality from the federal government. This document protects your study records from being subpoenaed (released to the courts at their request), and we will only release your records for a subpoena if you ask us to in writing.**

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As with the main study, any information or sample data that is shared with other researchers or databases will be identified by a unique number and not by your name or any identifying information unless you specifically give permission.

Also, if you choose to participate in the NUgene Project, you will have the option of being contacted in the future about other research opportunities as well as the option of receiving a newsletter.

Options for Participating in the NUgene Project

Yes	No	
_____	_____	I **agree** to participate in the NUgene Project

If you agreed to participate in the NUgene Project, please indicate your preferences for being recontacted below:

1. You have the option of being contacted in the future to ask questions about your health or to ask you to participate in more research. You are not obligated to participate further, but given the option to decide at that time. It is your responsibility to notify the NUgene Project if your contact information changes.

Yes	No	
_____	_____	The NUgene Project may contact me in the future to see whether I am interested in participating in other research studies.

2. You have the option to receive periodic newsletters on current research or other general updates about the NUgene Project.

Yes	No	
_____	_____	The NUgene Project may periodically contact me with an informational newsletter.

Options for Being Interviewed

You have the option of being interviewed and/or receive a survey after receiving your results. **Not all participants will be contacted to be interviewed or surveyed, even if they agree below.** If you agree to be contacted and interviewed, the interview will be scheduled at your convenience and could last up to 1 hour. If contacted, you will have

the option to do the interview either over the phone or in person and you will be compensated with $50 for your time (either paid in person or mailed to you). The interview will be audio recorded. This recording will be transcribed and you will be identified by a study ID number and not your name. All audio recordings will be destroyed at the end of the study. Surveys will be either mailed or emailed and will not collect any identifying information.

Yes	**No**	
_____	_____	I **agree** to be contacted to be interviewed & audio recorded and/or receive a survey

Signature Block for Capable Adult for the Main Study

Your signature documents your permission to take part in this research.

Signature of participant	Date
Printed name of participant	
Signature of person obtaining consent	Date
Printed name of person obtaining consent	