

## Permission to Take Part in a Human Research Study



*Title of research study: AdventHealth Orlando Genomic Population Health Study*

*Investigator: Rebecca Essner, PhD  
301 E Princeton Street  
Orlando, FL 32804*

**Daytime Phone Number:** 407-303-2800 x1107029

*Sponsor: AdventHealth Orlando*

Please check out our consent video for information about our study

Please be sure you have **not** had any of the following to ensure sequencing of your sample:

- Previous bone marrow transplant
- Blood transfusion in the last month
- Chemotherapy in the last 3 months
- Active diagnosis of hematological (blood) cancer

If you have any questions, please wait to sign the consent document until you speak with a coordinator at your scheduled appointment or you can email your questions to a member of the study team at [WholeMeFlorida@AdventHealth.com](mailto:WholeMeFlorida@AdventHealth.com).

### ***Research Consent Summary***

You are being invited to take part in a research study conducted by AdventHealth Orlando in collaboration with Helix OpCo, LLC (“Helix”), a genetics company who performs the sequencing of DNA, and companies working with AdventHealth Orlando and Helix (such as Admera Health, PerkinElmer Genomics, and Invitae) that provide genetic interpretation and confirmatory testing/genetic counseling services as part of this study. Your participation is voluntary. It’s your choice whether to participate. This summary describes the key information that we believe most people need to decide whether to take part in this research. Later sections of this document will provide all relevant details.

1. One purpose of this research is to provide information to research participants on whether they test positive for familial hypercholesterolemia (FH). This research will also include surveys to

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see how getting this genetic information affects research participants' health care practices and needs.

2. Another purpose of this research is to collect genetic information, medical record information, and insurance and billing information about research participants to create a database of information to use for other research in the future. The information in this database will not identify you, although there will be a way to connect the information in the database with you, in case future research shows that you are at risk for other genetic conditions.
3. There are some risks to participating in this research. You could find out that you or your relatives are at a higher risk of developing certain illnesses. This could create anxiety or stress.
4. Your genetic information, health information and survey responses will be stored in a database for the research study. While we have good security processes in place, in the unlikely event there is an unintended disclosure of information that can be used to identify you, it could affect your insurability, family relationships or other legal rights. You should know that there are laws in place to prevent this from taking place.
5. There are also benefits to participating in this research. The genetic testing done in this research could reveal information about you that is important for your health and the health of your relatives. For people with undiagnosed FH, taking action on this information could reduce your cardiovascular risk.

### ***Introduction***

Before you agree to be in the study, read this whole form carefully. It explains why we are doing the study; what you must do to participate in the study; what personal information, including genetic information and health information, you agree to share as a participant in the study; and other important information you need to know.

### ***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 ask all the questions you want before you decide whether you want to take part in this research.
- You can choose not to take part. There will be no penalty or loss of benefits to which you are otherwise entitled.
- You can agree to take part and later change your mind. There will be no penalty or loss of benefits to which you are otherwise entitled.
- If you are an employee of AdventHealth Orlando, your decision on whether to take part will not affect your employment or relationship with AdventHealth Orlando. It is completely voluntary.

### ***Why is this research being done?***

Genes affect our outward appearance, our underlying health or illness, and how we interact with our environment. Genes also tie us with our ancestors and where we come from. The American College of Medical Genetics and Genomics (ACMG) created a list of genes linked to a number of clinical conditions, where early diagnosis could help prevent or reduce bad effects from the condition (which

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we call “medically actionable genes”). Research, such as this, can contribute to this list of medically actionable genes.

The current list of medically actionable genes includes genes linked to clinical conditions such as heart disease, cancer, and other conditions that you can inherit from your parents. One of these conditions is familial hypercholesterolemia (FH), a common cardiovascular condition related to four genes that can explain this disease in about 80% of people. FH is often only diagnosed after a major heart event, but genetic testing could dramatically impact health outcomes if medical care is pursued following testing.

Through this study, AdventHealth Orlando researchers plan to:

1. Obtain demographic information and the saliva sample from Central Florida residents participating in this research study.
2. Provide the saliva sample to Helix for genetic testing. The genetic information provided to AdventHealth Orlando by Helix will initially include 60 medically actionable genes. There is also the possibility of AdventHealth Orlando getting genetic information for all known genes in the future.
3. Create a genetic and health information database without direct identifiers that will be used to look for genetic variation patterns to find genes that may be good for predicting and improving health.
4. Use this genetic information to discover relationships between genes, disease, and disease risk.
5. Inform research participants whether they are at-risk for FH or carriers (meaning they only have one copy of a gene affected) based on the genetic markers known at the time their genetic interpretation is performed by Helix’s genetic interpretation partners.
6. For those at-risk for FH or carriers, make genetic counseling available from Invitae, or other counseling groups used by Helix, to research participants.
7. Determine how the information on FH genetic risk changes or helps participants’ health care decision making.

### ***What happens if I agree to be in this research?***

You will first be asked to complete a brief intake form providing demographic information and a brief medical history. Then, you will be asked to create a Helix account, so that Helix can communicate with you about your genetic test results. When you create the Helix account, you will give them your name, date of birth, and your contact information. Helix will follow the process outlined in this consent form in how they handle your genetic information and the results of your genetic testing for this study.

When you create a Helix account, Helix will also ask you to consent to Helix using your information separately for research. This is a separate research consent form from this one and is also voluntary for you. You do not have to agree to the Helix research consent form in order to participate in this study.

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You will then be asked to provide a saliva sample. AdventHealth Orlando will send this saliva sample to Helix to do genetic testing on it. Helix will do genetic testing called “exome+ sequencing.” This is a laboratory process that maps out your genetic information, to identify if you have medically actionable genes. Helix will share the results of your genetic testing with AdventHealth Orlando. Helix will also share the results of your genetic testing that are related to FH with their partnered genetic interpretation lab, which will review the test results to see if you are at risk for FH. Four to eight weeks after Helix receives your sample, you will receive an email from Helix with information about your genetic results for FH.

Helix on behalf of its partner will also share its interpretation of your genetic test results with AdventHealth Orlando. If you are at risk for FH or a carrier, an AdventHealth Orlando care navigator will reach out to you to set up an appointment with a genetic counselor. Advent Health Orlando uses a company named Invitae (but a different company could be used in the future). The genetic counseling company will call you to discuss your genetic testing results.

The care navigator at AdventHealth Orlando will also set up an appointment for you at AdventHealth Orlando to get a blood test to confirm the positive result for FH. The care navigator will also make an appointment for you at the AdventHealth Orlando cardiovascular clinic. You do not have to agree to either of these appointments. If you do attend these appointments, the results of this confirmatory blood test and any care you receive at AdventHealth will be included in your AdventHealth medical record.

Next, researchers at AdventHealth Orlando will ask you to respond to an email survey (at 1 and 6 months after you receive your FH genetic results). This survey will ask you questions about any healthcare or lifestyle changes you have made as a result of getting information on FH. Your survey responses will not become part of your medical record.

Finally, AdventHealth Orlando researchers will collect information from your medical records and insurance and billing information that is relevant to the research. This may include your demographic information (such as your name, date of birth, and contact information), your medical history, a list of your present medical problems, medications that have been prescribed to you, your lab results, and other information from your medical record. By signing this form, you will be authorizing AdventHealth Orlando and your other health care providers to use and disclose your medical record information for this research. This includes information from your past, present, and future medical records. The next section describes who will have access to your medical record information and for what purpose.

### ***What happens to the information collected for the research?***

The following types of information about you will be collected for research:

- Demographic information.

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- Genetic information from Helix.
- Interpretation of your genetic test results. Helix will provide the interpretation for FH on behalf of their partnered genetic interpretation lab, but other companies may provide interpretation of genetic test results for other conditions.
- Summary report from genetic counseling session, if applicable
- Survey responses.
- Information from your past, current and future medical records. The type of information that will be collected from your medical records is described above.

This information will be used in two ways. First, this information will be used for the part of the study that tells you if you are at risk for FH based on your genetic test results. It will also be used to study how having this information about your risk for FH changed your health care decision making. For this part of the study, the following organizations and people may have access to information about you:

- AdventHealth Orlando
- Helix
- Helix's partnered genetic interpretation lab, such as Admera Health or PerkinElmer Genomics (or other companies that will interpret genetic test results)
- Invitae (or other genetic counseling companies)

Second, this information will be stored in a research database at AdventHealth Orlando to use for other types of research in the future. This information will be stored permanently in the research database, unless you ask us to stop. All of the information in the research database will have any information that directly identifies you removed. This de-identification process will be performed through a gatekeeper (which we call an "Honest Broker"). The gatekeeper will have the code that links you to the information about you that is stored in the database. The Honest Broker will not give this code to researchers.

If results from research in the future may be helpful for clinical treatment or early detection of diseases, the Honest Broker will be able to link the research information to you so that those research results may be returned to you. AdventHealth Orlando may give your genetic information to a company that interprets genetic test results. If AdventHealth Orlando returns research results to you in the future, AdventHealth Orlando may also give your information to a company that provides genetic counseling services.

The de-identified information in the database may be made available to researchers with AdventHealth Orlando, and to researchers at other organizations that are approved by AdventHealth Orlando. This could include researchers at commercial and for-profit companies. The information in the research database will be available for any type of research, such as research to understand what causes certain diseases (for example heart disease, cancer, or psychiatric disorders), development of new scientific methods, and development of new treatments for certain diseases. This research will be done without any additional consent from you.

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These researchers and organizations may also put your information into other scientific databases, where it will be combined with information collected from other research projects. Researchers can then study the combined information to learn even more about health and disease. For example, the National Institutes of Health (the NIH) maintains a scientific database. A researcher who wants to see the information in the scientific database must first get approval from the NIH. Your name and other information that could identify you will not be placed into these other scientific databases.

Information that directly identifies you will not be released to researchers without the approval of an Institutional Review Board (the IRB), a group of people whose role it is to protect people participating in research. In addition to the IRB, the Food and Drug Administration, the Office for Human Research Protections, the National Institutes of Health, US and foreign agencies, data safety monitoring boards, and accreditation organizations that regulate research may have access to information if necessary. Your information may also be disclosed if required by law.

### ***How much time will this take to participate in the research?***

The study enrollment will take about 60 minutes of your time. This will include the education and registration process and providing a saliva sample.

One and six months after you receive your FH genetic results, you will be contacted by AdventHealth Orlando researchers via email and asked to answer several questions about what health care actions you took after receiving your genetic results for FH. These surveys will only take a few minutes to complete.

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

Taking part in this research involves no potential harmful consequences to your health. However, there are risks involved in having your genes analyzed and in sharing your genetic and health information.

- Your genetic data may reveal that you are at a higher risk of developing certain illnesses, which might also indicate that your genetic relatives are also at-risk. Finding out you have a higher risk of certain conditions, such as cancer or heart disease, can create stress or anxiety.
- People whose genetic testing does not show a genetic condition may think they are healthy. Genetic testing is not perfect and there is a very small chance that the genetic test does not show a problem, but the person actually has a genetic abnormality.
- People whose genetic testing does not show a genetic condition may think they are healthy and could ignore other potential health risks. Approximately 20% of FH is not a result of any current DNA test available today.

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- People whose genetic testing does not show a genetic condition could still have FH. Certain types of genetic changes within this panel of genes may not be detected with this method, which accounts for approximately 5-10% of FH cases.
- Some survey questions may make you or your family members uncomfortable.
- If you or a family member has genetic data linked to your name or your family member's name in a public database, someone who has access to your de-identified genetic data might be able to link that data to your name or your family member's name through the publicly available genetic data.
- Your genetic information, health information and survey responses will be stored in a database for the research study. (See more information on that above.) AdventHealth Orlando has strong policies and procedures in place to reduce the possibility of your information being stolen or lost, but they cannot provide a 100% guarantee that your information will be safe. In the unlikely event there is an unintended disclosure of information that can be used to identify you, it could affect your insurability, family relationships or other legal rights. You should know that there are laws in place to prevent this from taking place. A federal law called the Genetic Information Nondiscrimination Act (GINA) makes it illegal for health insurance companies, group health plans, and most employers to discriminate against you based on your genetic information. GINA does not apply to life insurance, long term care insurance, or disability insurance. Please ask the study investigator or study staff if you would like to know more about how your information will be protected while you are in this study.
- Your information may be provided to outside researchers. Once your information is given to outside researchers, it is possible that your information will no longer be protected by the federal privacy law called HIPAA.

### ***Will being in this study help me in any way?***

The report on your genetic information could reveal information about you that is important for your health and the health of your relatives. For this initial phase, markers for FH are rare in the general public (that is, those without a family history of coronary artery disease) so the chance of finding a positive result is small. For people with FH, taking action on this information could reduce your cardiovascular risk. You may benefit from sharing the report with your healthcare professional who can help you better understand the genetic information contained in the report.

You will receive your FH test results and an explanation of those results. You will receive notice that your FH results are available through an email from Helix. These results and an explanation of the results will be available in the FH report from Helix. More information about the Helix genetic test and test results will be provided to you when you set up an account on the Helix website.

An AdventHealth Orlando care navigator will also be available to direct you to physicians and other resources for help.

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Your genetic information will be stored in the AdventHealth database and will be used to analyze and interpret genetic information associated with other conditions that are inherited through families, such as heart disease and cancer. As a participant in this study, you may receive information about your genetics, which could result in getting early screening, early detection, and early treatment of conditions. Please keep in mind that the science about genes is changing quickly, so you should not rely on AdventHealth to tell you about your risk of genetic conditions. You should continue to live in a healthy way and seek preventive care from your health care providers.

Also, when you create an account on the Helix website, you will have access to other Helix store products related to other health conditions. These are available at an additional cost to you but require no additional saliva sample. Results from these purchases will not be shared with AdventHealth Orlando. As part of this research study, you will receive two products from the Helix Store at no charge to you, which includes entertainment traits (both an ancestry insights product and non-clinical product with traits such as lactose and gluten tolerance).

### ***Are there any costs in this study?***

There is no cost to you associated with participation in this study. You do not need to pay for Helix's genetic testing.

If you are contacted with a positive finding for FH, you will not have to pay for the blood test to confirm whether you have FH, your initial cardiologist visit, or your initial genetic counseling session. However, you and your health insurance company will be responsible for any other follow up care you choose to receive.

If any genetic testing results about other conditions are given to you in the future, any follow-up care, including confirmatory testing, genetic counseling sessions and additional procedures, will be your responsibility.

### ***What other choices do I have besides taking part in the research?***

You can decide not to be in the study. There are other companies that will do genetic testing for you for a fee. You may not participate in this research study if you do not sign this permission form.

### ***How many people will be studied?***

We expect about 9250 people will take part in the entire study.

### ***What else do I need to know?***

The research done with your medical information and genetic information may help develop new medical products in the future. There are no plans for you to share in any financial benefit resulting from the research or the development of these products, and you will have no property or other interest in the research or these products.



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### **May I cancel my permission to take part in this research later?**

Yes, you may cancel your permission to participate in this research at any time. You do this by sending written notice to the study investigator:

***Rebecca Essner, PhD  
301 E Princeton Street  
Orlando, FL 32804***

When you cancel your permission, no new information about you will be gathered. Information that has already been gathered may still be used to support the FH research study. Your study cancellation will not affect your relationships with Helix or Admera Health.

Information in the AdventHealth Orlando database is de-identified and will stay in the database for future research. However, if you cancel your permission to participate, we will destroy the link from the database to your identity. If that happens, we will not be able to notify you about any future research results about an identified genetic condition.

This permission will not end unless you cancel it in writing.

Your participation may also be cancelled in the unlikely event that Helix is unable to sequence your saliva sample. Helix will ask for a second sample by mailing another kit directly to you. After 2 unsuccessful attempts, you will be withdrawn from the study.

### ***Who can I talk to?***

If you have questions, concerns, or complaints (or you decide to participate and later think the research has hurt you), you can call the research team at the phone number listed above on the first page. This research is being overseen by an Institutional Review Board (“IRB”). The IRB is a group of people who review and approve research at AdventHealth Orlando. You may talk to them at (407) 200-2677 or [FH.IRB.General@adventhealth.com](mailto:FH.IRB.General@adventhealth.com) if:

- Your questions, concerns, or complaints are not being answered by the research team.
- You are not getting answers from 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 subject.
- You want to get information or provide input about this research.

You also have a right to request a copy of the AdventHealth Orlando Privacy Notice. If you have questions about your privacy rights, you may contact AdventHealth Orlando’s Privacy Officer at PH: (407) 200-2961.

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### Signature Block for Adult Subject Able to Consent

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