

**University of North Carolina at Chapel Hill
Consent to Participate in a Research Study
Consent Form 1: Randomization to Research Genomic Sequencing
Child Participants
Biomedical Form**

IRB Study # 17-1806

Consent Form Version Date: 02-28-2018

Title of Study: North Carolina Clinical Genomic Evaluation by Next-gen Exome Sequencing, phase 2 (NCGENES 2)

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This consent form is for participants who have already enrolled in the first part of the NCGENES 2 study and who are being invited to join the second part of the study.

What are some general things you should know about research?

Research is done to gain new information that may help other people in the future. You and your child *may not* receive any direct benefit from being in this research study. There may also be unknown risks. You may refuse for your child to participate in this research. If your child is a patient with an illness, he or she does not have to be in this research study to get medical care.

Details are discussed below. It is important that you understand this information so that you can make an informed choice. You will be given a copy of this consent form. You should ask the researchers or research staff any questions you have about this research study at any time.

What is the purpose of the NCGENES study?

This part of the NCGENES study is looking at the effect of using a genetic test called “genomic sequencing.” We want to learn whether genomic sequencing can diagnose a medical condition sooner and change a child’s future medical care. We are comparing it to what happens when children get usual care. “Usual care” includes all the evaluations and tests done as part of a specialty clinic visit. Right now, genomic sequencing is not often part of usual care.

We also want to learn how families understand, talk about, and use information they get from their child’s specialty clinic visits.

How many people will take part in this study?

We expect about 850 families will join this part of the study.

What will happen if you and your child join this part of the study?

We will ask you to sign a HIPAA form so we can access your child's medical records to obtain health-related information from his or her visits to UNC Hospitals. We will only access information that is needed for this study. This information includes your child's health history, family medical history, and some laboratory test results.

We will use "randomization" to divide the families who join this part of the study into two groups: Group 1 and Group 2. Randomization is like flipping a coin. It makes sure that every family has an equal chance of getting in one group or the other. You will be told which group you are in after you consent to being in this part of the study and sign this consent form.

If you are in Group 1:

Children will get usual care from their specialty clinic doctors. This is the same care that any child would get in this clinic if they were not in the NCGENES study. It includes a physical exam and it *may* also include one or more of the following;

- Additional testing, including some kinds of genetic tests
- Medical procedures, such as an MRI
- Referral to other specialists, including therapists
- Other recommendations

These things are all part of usual care. They are not part of the NCGENES study and the study will not pay for them. However, we will track their results to see if they affect your child's future health and medical care.

If you are in Group 2:

Children will get usual care from their specialty clinic doctors, as described above. They will *also* be offered genomic sequencing to look for a genetic reason for their condition.

If you are in Group 1:

You do not have to do anything.

If you are in Group 2:

We will give you more information about genomic sequencing. We will then ask you to decide whether you want genomic sequencing for your child. To agree to this testing, you will sign a separate consent form. You can also decide you do not want genomic sequencing for your child. Your choice will not affect your child's medical care.

How long will your and your child's part in this study last?

Your child will probably be scheduled for a routine follow-up clinic visit about 6 to 12 months after his or her first clinic visit. At that time, the doctors will discuss test results from your child's usual care and from your child's genomic sequencing, if it was done. In some families, this follow-up contact might be done by phone.

After the follow-up contact, we will check on your child's health over time, including after the study has ended, by using his/her medical records. We may also check on your child's health by looking at other large data sets such as health insurance claims data. We will stop doing these

things when your child turns 18 years old. Additionally, we will contact you if we find new information during the study that would affect your child's medical care or your willingness to continue in the study. We may also contact you about joining future research studies. You can accept or decline to join these studies at that time.

What are the possible benefits to you?

The goal of research is to benefit society by gaining new knowledge. The study will help us understand how to use genomic sequencing with patients and help us find out if that use affects a child's future medical care or not.

What are the possible risks involved with participation in this study?

Risk to Confidentiality and Privacy: There is a small risk to your privacy and the confidentiality of the information we gather in the study. However, this study has many ways to protect your privacy and confidentiality. These include using participant ID numbers, special ways to store information, and how we share our research findings.

Use of Participant ID Numbers: All study materials will be coded with a unique participant ID number instead of your name or other information that can be used to identify you or your child. We will store the records that link your ID number to your personal identifying information on a secure computer in a password-protected file with restricted access.

Electronic information: Information, including information from your child's medical records, will be stored on secure computers in password-protected file with restricted access.

Paper Documents: Paper documents will be stored in a locked filing cabinet in locked offices.

Research Publications: When we report results from this research study, we will not identify you or your child in any published reports

We will make every effort to keep your research records private. However, there may be times when federal or state law requires us to disclose them. This is very unlikely, but if disclosure is ever required, UNC-Chapel Hill will take steps allowable by law to protect the privacy of your personal information. In some cases, the information could be reviewed by University representatives or research sponsors for quality control or safety.

To help us protect your privacy, we have obtained a **Certificate of Confidentiality** from the National Institutes of Health. That means we cannot be forced to disclose information that may identify you, even by a court subpoena, in any federal, state, or local civil, criminal, administrative, legislative, or other proceedings. We will use the Certificate to resist any demands for information that would identify you, except as explained below. The Certificate cannot be used to resist a demand for information from personnel of the United States Government if they need to audit or evaluate a federally-funded project or if it is required by the federal Food and Drug Administration (FDA).

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 you give an insurer, employer, or other person your written consent to receive research information, then we may not use the Certificate to withhold that information.

Other risks to study participation: There may be uncommon or other risks of being in this study that we currently do not know about. You should report any concerns to the researchers listed on the first page of this consent form.

Who is sponsoring and paying for this research?

This research study is being paid for by a grant from the National Human Genome Research Institute at the National Institutes of Health (NIH). We are paid to carry out the study but we do *not* have a direct financial interest with the sponsor or in the final study results.

Data Sharing with Qualified Researchers

By signing this consent form, you are allowing us to share information about your child with researchers to study the clinical use of genomic sequencing.

- Study data that is linked to your child's personal information (identifiable data) will be shared among NCGENES investigators through a secure, restricted-access data system and under a contract known as a Data Use Agreement that assures privacy protections.
- Coded study data, the data that does not include your child's personal identifying information, may also be shared with other investigators who are not members of NCGENES but who have been approved by the NCGENES steering committee.

The NIH is the government agency that pays for most medical research in the United States. Because this study is funded by NIH, we will share certain data with NIH and other data banks. The data we share will not include any personal identifying information. By collecting the health information obtained from many research centers, the NIH and other data banks will store it so other qualified researchers can use it to do more studies. Research can be done at an academic, government, and/or a commercial sites and studies may be done at many places at the same time.

Risks to Privacy and Confidentiality by Data Sharing

We think there are low risks to your privacy and confidentiality from sharing your child's study-related health information and questionnaire responses with other databanks; however, we cannot predict how this information will be used in the future. These databases have safeguards to protect information while it is stored and used for research. If your child has a genetic condition, information will be labeled with only a code number. His or her personal identifying information will *not* be used.

If you no longer want your child's data in these databases, you can withdraw your consent to participate in this study at any time with no penalty by contacting the researchers on the front page of this form. However, data that has already been sent to researchers cannot be retrieved from them.

Will researchers ask your permission to do future studies involving the samples?

A committee called the Institutional Review Board (IRB) protects the rights and welfare of research participants in current and future research. They will decide about the need for future contact. In some cases, the IRB may determine that future research using your child's study-related data is acceptable without re-contacting you. In other cases, the IRB may require that you be re-contacted and asked for your consent. You have the right, at that future time, to refuse to allow your child to participate. This refusal will *not* affect your or your child's medical care, and it will not result in loss of benefits to which you are or your child is entitled.

Optional Consent for Storing Study Data with Identifying Information After the Study Ends:

In the future, after this study is over, researchers may identify new questions that might be answered with the use of your and your child's study data. The data from the NCGENES study will be stored in what we call a "repository" or "data bank". The kind of data that the study will store includes your answers to survey questions, information from your child's medical records that was used for NCGENES, or other related data. We will store this information labeled with only a code or number so that neither you nor your child are identified. We will not share data that has your or your child's name or personal identifying information on it. We will keep a list that can link you and your child's identity to the code, but we will not share this link with anyone outside of the study team. When your child turns 18 years old, we will remove the link so their information will be completely de-identified, that is no longer linkable to their identity.

It is unlikely that you will benefit from these future research studies. Future studies that use the data from this repository or data bank may help us develop new research projects and answer new research questions. For example, future studies may provide additional information that will help us understand the different types of conditions children have, how families search for answers to the cause of their child's health condition, how doctors communicate with families, and the types of tests and care that children who have these kinds of conditions receive throughout their lifetime.

The risks of participating in these future research studies are similar to the risks described above. Sometimes people are concerned that others may find out things about them or their child. For example, they may be worried that someone will find out about an answer to a survey question or information from a medical record. There is a low chance that others will find out about these things if the data are stored in this repository or data bank because the data will not be linked to your identity. Researchers who use these data will not know which data came from you or your child and which came from other people in this or other studies. There will be no cost to you for the storage and use of the data for research purposes. At any time, even after the study has ended, you can decide that you want to withdraw your and your child's data from the data bank. To do so, you should contact the researcher whose name is listed on the front page of this consent form.

Do you agree to the use of you and your child's data collected for the NCGENES study in future studies after NCGENES?

_____ I agree to this use

_____ I do **NOT** agree to this use

Can you withdraw from participation in this study?

Yes. You can withdraw from this study at any time, without penalty, by contacting the researchers listed on the front page of this form.

What if we learn new things or information during the study?

You will be given any new information we gain during the study that might affect your medical care or your willingness to continue participating.

What will happen if you are or your child is injured by this research?

All research involves a chance that something bad might happen to participants including the risk of personal injury. UNC-Chapel Hill has not set aside funds to pay for any such injuries, or for the related medical care. However, by signing this form, you do not give up any of you or your child’s legal rights.

Will there be any cost to you for participating in this research study?

You will not be charged for participating in this part of the study.

Will you receive anything for your participation?

We will not pay you nor your child for your participation in this part of the study.

What if you have questions about your rights as a research participant?

The IRB reviews all research on human volunteers to protect your rights and welfare. If you have questions or concerns about your rights as a research participant you may contact, the IRB at 919-966-3113 or to IRB_subjects@unc.edu. You do not have to use your name.

Participant Agreement:

I have read the information provided above and have asked all the questions I have at this time. I voluntarily agree to my and my child’s participation in **the North Carolina Clinical Genomic Evaluation by Next-gen Exome Sequencing, phase 2 (NCGENES 2)** study. **Principal Investigators:** Jonathan S. Berg, MD, PhD, Bradford Powell, M.D., PhD, Christine Rini, Ph.D.

Signature of Research Participant’s Parent or Guardian

Date

Printed Name of Research Participant’s Parent or Guardian

Relationship to Research Participant

Research Participant’s Printed Name

Signature of Research Team Member Obtaining Consent

Date

Printed Name of Research Team Member Obtaining Consent