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Main Study: Parent of Cognitively Intact 18-21 year old child

STUDY INFORMATION:

Study Title: TeleKidSeq: Incorporating Telehealth into the Clinical Care of Diverse NYC Children Undergoing Whole Genome Sequencing

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SUMMARY OF THIS RESEARCH STUDY:

In medicine there are many unanswered questions. A "research study" is when scientists try to answer a question about something that we don't know enough about. Participation in a research study may or may not directly help your child or others. Participation is entirely voluntary. It is completely up to you whether or not your child takes part. You can also change your mind at any time and it will not affect your child's ability to get medical care within the Mount Sinai Health System or elsewhere.

The purpose of this research study is to learn how genomic testing can help children and young adults with rare diseases. Genomic testing is a way for scientists to study your child's DNA (genetic material inherited from their parents that at least in part determines your child's features like eye color, height, and risk of many diseases). Sometimes, genes have changes, or "variants," that cause them to not function correctly, resulting in disease. These variants can be inherited from parents or can occur randomly. One type of genomic testing is called whole genome sequencing (WGS), which reads through all of a person's DNA. This consent form is focused only on genomic testing.

In this study, we will use WGS to try to learn the genetic cause of your child's condition. We will perform these tests in a clinically certified laboratory, and the results will be shared with you and your child's physician.

A major goal of this study is to learn the best way to communicate these complicated genomic results back to families like yours, by having parents answer a series of surveys. Everyone in the study must have a least one parent available to answer these surveys. As part of this study, all visits will be conducted using "telehealth," a way of delivering health services using communication technology, like video conferencing. Studying the use of telehealth for genetic testing will help healthcare providers understand how to improve patients' experiences in using communication technology. Additionally, we

hope to help scientists and healthcare systems learn how to offer and perform genomic testing to more people from diverse backgrounds and cultures.

If you choose to participate, you and your child will be asked to take part in three study visits. Each visit will last about 1 to 2 hours. You and your child's participation in this research study is expected to last nine months.

If you sign this consent form, you voluntarily agree that your blood, saliva or cheek swab sample and sequencing information can be stored indefinitely by the research study, including research teams at New York Genome Center, Einstein/Montefiore, and Mount Sinai. Samples may be used for either research or for clinical purposes if additional testing is needed. Your identifiable data may be used by the research team for reasons related to, and for reasons unrelated to, the current research project.

The main risks to you if you choose to participate are risks related to learning genetic information and the possibility of a loss of confidentiality or privacy. There is a chance that you may learn that your child carries a genetic change that may increase the risk for a specific medical condition. If that is the case, we may suggest that other members of the family get tested for the same genetic change, and you may learn that a family member is at risk to develop certain medical conditions or diseases. This knowledge might be upsetting and may cause you to have anxiety or psychological distress. Loss of privacy means having your personal information shared with someone who is not on the study team and was not supposed to see or know about your information. The study team plans to protect your privacy – see the Confidentiality section later in this consent form for details.

Your child may also benefit from participation in this research if WGS identifies a genetic diagnosis for your child. This information can inform your child's care and treatment and can be used for reproductive decision-making. In addition, understanding genetic diversity can help all people benefit from genomic medicine. Helping healthcare providers and scientists learn how we can best communicate information about WGS may help individuals who choose to have WGS in the future.

Instead of participating in this research, your child may be referred to Clinical Genetics for evaluation and to coordinate genetic testing.

If you are interested in learning more about this study, please continue to read below.

PARTICIPATION IN THIS RESEARCH STUDY:

This research study will be fully explained to you by a member of the study team. Feel free to ask all the questions you want before you make a decision about whether or not to participate. Any new information that develops during this research study that might make you change your mind about participating will be given to you promptly.

You may qualify to take part in this research study because your child is age 0-21 years and currently has an undiagnosed, likely genetic* cause of neurologic, immunologic, or cardiac disorder(s). If the child had genetic testing previously done, results must have been returned at least three months before enrollment and results must have been negative, or identified only one variant in a potentially causative autosomal recessive gene or variant(s) of uncertain significance. Participating parent(s)/guardian(s) must have access to the Internet and a device capable of videoconferencing via Zoom or be willing to use one that is provided in order to participate in this study.

Funds for conducting this research are provided by National Human Genome Research Institute (NHGRI) and the National Institute on Minority Health and Health Disparities (NIHMD) of the National Institutes of Health (NIH).

LENGTH OF TIME AND NUMBER OF PEOPLE EXPECTED TO PARTICIPATE:

Your participation in this research study is expected to last 9 months, which will involve three study visits conducted through telehealth. Each study visit will last about 1 to 2 hours.

The first two visits will be for you and your child. These two visits are held about 3-4 months apart.

The third (last) visit will be either a phone call or a video conference visit for you to answer a survey six months later.

The number of people expected to take part in this research study at Mount Sinai Health System is approximately 250 children and 375 parents, with another 250 children and 375 parents participating at the Montefiore Medical Center for a total of 1,250 participants.

DESCRIPTION OF WHAT'S INVOLVED:

If you agree to permit your child's participation in this research study, the following information describes what may be involved.

Baseline Study visit: Initial genetic counseling visit (1-2 hours)
At this first visit, you will meet with a genetic counselor via telehealth. This allows for the completion of this study visit in your own home. All participants will receive genetic counseling using the usual care that is normally given. The telehealth platform (Zoom) used provides video and audio support, which will allow you and the genetic counselor to see and hear each other. The Zoom link information was emailed to you beforehand along with instructions on how to set-up and use Zoom. Half of the study participants will *also* use a screen-sharing capability on the telehealth platform while the other half will not. This allows the genetic counselor to present images to you on the screen. Assignment to telehealth with screen-sharing capability or no screen-sharing will be chosen by chance, like flipping a coin. Your chance of being in either group is 50:50. Neither you nor the study doctor will choose what experimental study treatment your child gets. Your child will have a(n) equal chance of being given each experimental

treatment. Your random assignment to either group occurred before your first study visit and you will be told of your assignment at the end of your first visit with the study genetic counselor.

The genetic counselor will ask you a very detailed family medical history, including grandparents, parents, children, aunts, uncles, and cousins on both sides of the family.

At the same visit, the genetic counselor will ask about your child's medical history including the medicines they are taking. We will share the family history and medical history with the testing laboratory as it may help with the interpretation of your child's genomic results.

During this initial counseling session, the genetic counselor will also explain DNA and genes to you, and will go over the types of information we might learn from your child's genetic testing.
Since family members share genetic information, the information from this test may apply to your family members, as well. The genetic counselor will talk you through this information and how the findings may affect you and your family.

If you do not have access to the internet and/or an electronic device capable of using a teleconferencing platform, the study staff will help to make arrangements for you to complete the Baseline study visit study visit at a Mount Sinai facility or your provider's office. The visit will occur in a secure space using a tablet or computer provided by the study staff. No visits will be conducted face-to-face with the study staff, as an aim of this research is to study the efficiency of telehealth.

Your child will receive whole genome sequencing (WGS), a test that will look for genetic changes that might be causing your child's epilepsy, developmental delays, heart disease, or low immune system. This test will be done at the New York Genome Center's clinical laboratory (NYGC).

Whole genome sequencing, or WGS, is a genetic test that involves sequencing, or reading through, all of a person's DNA. Your child's genome will be sequenced and compared with the DNA from individual(s) that do not have any known disease. There are millions of differences, or variants, between people that cause us to each be unique. All of the differences will be studied by the laboratory scientists to see which, if any, are related to your child's condition. Besides the possible disease-causing variants and the "secondary findings" variants described below (if you chose to receive them), no other variants will be reported, even if there are variants that show that your child has or is at risk for an unrelated genetic disease. Because interpretation of the WGS is dependent on complete and accurate medical records, your physician and the genetic counselor will provide detailed medical information to the NYGC lab. Although WGS is the most thorough type of genetic testing currently available, it is a relatively new test. In addition, scientists are rapidly learning about the human genome, but there is still a lot that we do not currently understand. For those reasons, it is possible that disease-causing variants may be missed on your child's WGS. The types of possible results from WGS testing are listed below.

NYGC is a CLIA and CAP certified laboratory. Being CLIA certified and CAP accredited ensures your test results are meeting and exceeding industry standards for clinical laboratory testing.
The types of changes that we *might* find in your child's DNA include (but are not limited to) the

following:

1. Positive results: We might find abnormal changes (or "pathogenic variants") in a known disease gene that explains why your child has health problems. This will give us a diagnosis for your child's condition.
2. Likely positive results: We might find variants in a known disease gene that have NOT been seen in other people with the same disorder or have been observed in very few individuals, but that are *likely* causing your child's health problems.
3. Uncertain results: We might find genetic changes (or "variants of uncertain significance") that are inconclusive or uncertain. Sometimes we find a genetic variant that has not been seen before, and we do not know if it is disease-causing or just a difference that can exist without causing disease.
4. Negative results: We might have a negative result, where we do not find genetic variants related to your child's condition.
5. We might find "secondary findings," or variants in genes that are NOT related to your child's disorder, but that might be important for their health. These are a specific set of genes recommended by the American College of Medical Genetics and Genomics (ACMG). Many of these genes involve inherited forms of heart disease and cancer, and when a person has variants in these genes, it puts them at higher risk to develop the disease. If we find that your child has a disease-causing variant in one of these "secondary findings" genes, we will tell you about it and refer you for appropriate care. Importantly, if you are positive for disease-causing variants in a "secondary findings" gene, there are other relatives, including yourself and any other children, who may have the same disease-causing variant and who therefore may be at risk to have a disorder. If your child tests positive, therefore, we will recommend testing family members. Your child will have the option to choose whether he/she would like to receive information about his/her secondary findings.

Sample collection (30mins)
At the end of the genetic counseling session, the genetic counselor will provide directions on how to collect a saliva (about 1 teaspoon) or cheek swab sample from your child and from each biological parent (if available). Sample collection kits will be provided to you with pre-paid return mailing packages. The genetic counselor will walk you through the steps of sample collection during that first visit. Upon completion of the sample collection, you will package the samples according to the directions and place the package in the mail. It will be received either by the study team or by the laboratory (NYGC).

There is also the option to take a sample of your child's blood if for any reason you are unable to collect a saliva or cheek swab sample from them. This will require an in-person visit. This will occur at a Mount Sinai facility or your provider's office. A trained medical professional will wipe the skin on your child's arm with alcohol to clean it. Then, he/she will insert a small needle into a vein and 2 tubes of blood will be drawn, about 10-40 ml of blood (1-3 Tablespoons). We would also like to take about 3 tablespoons of blood from each biological parent (if available).

We will share your child's name, date of birth and medical record number on the laboratory test order that is sent along with your child's samples, as well as copy of this consent and your child's detailed medical and family history. The clinical laboratory needs this information in order to report the results of your child's test and for it to be a part of your child's permanent medical record.

Parental samples will only be used to help us understand your child's DNA results. For example, if your child has a variant, we might use your blood to see if it was inherited from a parent. It is completely voluntary for parents to give samples. Your child may take part in this study without parental samples, but having them increases the chance of identifying the genetic cause of your child's condition and decreases the chance of uncertain or unclear results.

Because we are only analyzing parent samples to understand your child's results we will not look for or find genetic changes that cause other diseases for parents. Your names will not appear in the reports and you will not have separate results or reports. However, this test may suggest that biological relationships of family members are not as reported, such as non-paternity (the man identified as the father of the child is not the biological father). The lab report will not directly state that there is a question about paternity, but people reading the report may be able to figure it out nonetheless. These samples will be sent to the study lab along with your child's sample. If we are unable to receive a sample from you (or your child's other parent), you can come back at a later date to have the sample taken, or we may provide you with a kit to collect saliva or cells from the inside of your cheek.

After receipt of the child's sample, the study team will give you a $20 gift card by mail or email for Study Visit 1.

Study Visit 2: Return of Results (ROR) with a genetic counselor (1-2 hours)
After your child's DNA is read and interpreted, it will be reported to the study's medical geneticist, the physician who ordered the test, and your genetic counselor. This will occur about three months after your first visit. You will be asked to attend a genetic counseling session via telehealth to review the results in detail. This allows for the visit to be completed in your own home.

If you do not have access to the internet and/or an electronic device capable of using a teleconferencing platform, the study staff will help to make arrangements for you to complete this study visit at a Mount Sinai facility or your provider's office. The visit will occur in a secure space using a tablet or computer provided by the study.

If a diagnosis was made, it will be important for you to notify all of your child's doctors of the diagnosis. Your child's genetic testing results will be stored in his/her permanent medical record. This report will be limited to genetic results related to your child's diagnosis, and secondary findings if you chose to learn about them.

All participants will receive their genetic results using the usual care that is normally given at a return of results session. However, if you were randomized to the arm of the study that allows for screen-sharing,

this telehealth capability will also be used. At the end of this visit, you will complete the return of results survey described above. This survey will help us see if screen-sharing capabilities were effective at helping people understand their results in telehealth compared with genetic counseling in telehealth that does not include screen-sharing capabilities. We will give you a $20 gift card for this visit.

Study Visit 3: Assessing your understanding of your results by survey (1 hour)
About nine months after your first visit and six months after your return of results session, you will have another study visit. The point of this visit is to see how well you understood your child's results, a measure for us to see how well we communicated the results to you. This visit will involve completing the third and final survey, and can be done either in person or over the phone. We will give you a $40 gift card once this last survey is completed.

After the Study Visits: Reviews of your DNA and medical records
We will analyze the costs associated with any symptoms or events your child experiences through the follow-up period. The researchers may collect billing information from your child's hospitalization stays and treatments outside the hospital.

COVID-19 procedures related to in-person activities
For in-person activities related to the study (ie, blood draw), the following will occur:

The research team will contact you within 24 hours of visit(s) to conduct pre-visit screening for COVID-related symptoms using the most up-to-date MSHS Infectious Diseases Screening Tool.

You and your child will be asked to come alone and to wear a mask to the study visit. If you do not have one, one will be provided to you upon arrival by study staff. All hospital/departmental/clinic rules regarding COVID-19 prevention will be followed once you arrive on-site, including but not limited to pre-visit screening at an established ambulatory practice area, wearing a mask at all times, etc.

The study team has put in place several procedures to minimize exposure to COVID-19, including using masks, eye shields, and gloves and practicing social distancing. Study staff screen for COVID-related symptoms daily before their work shift begins to ensure they are fit to engage in person with study participants. The study staff will try to minimize the time you need to be on site to complete a study visit, and will complete as many study procedures as possible via telehealth.

USE OF YOUR DATA AND/OR SPECIMENS:

Storage and use of your leftover blood sample and data within TeleKidSeq

By signing this consent form, you voluntarily agree that you and your child's blood and sequencing information can be stored indefinitely by the research study, including TeleKidSeq research teams at New York Genome Center, Einstein Montefiore, and Mount Sinai. Samples may be used for either research or for clinical purposes if additional testing is needed. Your child's identifiable data may be used by the TeleKidSeq research team for reasons related to, and for reasons unrelated to, the current

research project. If you decide that you do not want the TeleKidSeq research teams to keep your or your child's biological samples, you may withdraw your consent to storage and to use of your samples at any time by contacting Dr. Eimear Kenny (contact information on first page of consent), in which case we will promptly destroy the sample(s) or the portions thereof that have not already been used. However, you or your child's sample may have already been distributed to other researchers within NYCKidSeq before you ask us to destroy it, so we may not be able to retrieve it and stop future research.

To protect your privacy, Mount Sinai has policies and procedures in place that are overseen and monitored by Institutional Review Board. Mount Sinai Health System requires its staff who may use or have access to your or your child's samples or data to receive training on its privacy and data security policies, and to follow those policies with care.

Sharing your leftover sample and data with researchers outside of TeleKidSeq

We would like to ask your permission to store and share your blood, saliva and DNA samples, and sequencing information (data), which will be stripped of identifiers to protect your confidentiality, with other researchers (i.e. those who are not associated with TeleKidSeq, Einstein Montefiore, NYGC, Mount Sinai). These biological samples and the sequencing data may be used in future research, including in future genetic testing, to learn about, prevent, or treat health problems.

> **You must initial your choice. By initialing you are consenting to the following:** TeleKidSeq has my permission to store my leftover sample and to share my de-identified data and/or sample with researchers outside of TeleKidSeq.
>
> _____(Initial) Biological Mother
>
> _____(Initial) Biological Father

Public Sharing of your genome data

One purpose of this study is to help researchers around the world learn about the genomes of people from diverse populations. They do this by putting it into one or more scientific databases, where it is stored along with information from other studies. Researchers can then study the combined information to learn even more about health and disease.

If you agree to share de-identified data in secure, public research databases, some of your child's genetic and related health information will be entered into one or more scientific databases available to other researchers inside and outside of Einstein-Montefiore, Mount Sinai, and the New York Genome Center. For example, the National Institutes of Health (an agency of the federal government) maintains a database called The Database of Genes and Phenotypes ("dbGAP"). A researcher who wants to study the information must apply to the database. Different databases may have different ways of reviewing such requests. However, only researchers who apply and are approved can access restricted databases, like dbGAP, dbVar, and other databases. The TeleKidSeq program will limit sharing of individual data to only those restricted databases, which require approval to access.

Please note that identifying information about your child, such as your name, address, telephone number, or social security number, will NOT be put into these scientific databases. However, because your genetic information is unique to you, there is a chance that it could be traced back to you. The risk

of this happening is very small and is explained in the Risks section of this consent form. Researchers will always have a duty to protect your privacy and to keep your information confidential.

You must initial your choice. By initialing you are consenting to the following: TeleKidSeq has my permission to store and deposit my de-identified clinical information and sequencing data in secure, public research databases.

_____(Initial) Biological Mother

_____(Initial) Biological Father

Participating in future research studies

As new research opportunities are identified, or new research findings made, the researchers may wish to contact you to ask if you would be willing to donate fresh samples for additional testing, or to share information about research progress with you, or to invite you to enroll in new studies.

However, this is not a requirement to take part in this study. A separate consent will be obtained if you wish to take part in future research.

If the researchers are aware of a research project that might be relevant to your child, do you give them permission to contact you in the future to collect additional information about your child, share information with you, or to discuss possible participation in another research project?

You must initial your choice:

_____(Initial) I consent to be contacted in the future to learn about new research studies that my child may wish to join or new research findings.

_____(Initial) I consent to be contacted in the future if the researchers would like additional samples from me or my child.

_____(Initial) I do NOT want to be contacted by researchers seeking to collect or share additional information or to discuss another research project.

YOUR RESPONSIBILITIES IF YOU TAKE PART IN THIS RESEARCH:

If you decide to take part in this research study you will be responsible for the following things:

- Return for your follow-up visits and complete your study surveys.
- Collect a saliva or cheek swab sample from your child and each biological parent (if available) and send them back with the pre-paid packaging label

- If you think you are pregnant or fathering a child, please let your research study team know. If we find that your child has a genetic variant that is causing his/her disease, there is a chance that the same variant may affect another pregnancy. If you tell us that you or your child is pregnant or fathering a child, our genetic counselors will discuss this with you in detail.

COSTS OR PAYMENTS THAT MAY RESULT FROM PARTICIPATION:

The genetic counseling sessions and WGS will be provided to you at no cost. Taking part in this research study may lead to added costs to you or your child. Should you choose to participate in study visits in person in a hospital setting using telehealth, you and your child will not be reimbursed for your or your child's travel or time that may be required for study visits. Depending on the results of WGS, further testing, screening, and/or procedures may be recommended as part of your child's or other family members' clinical care. Costs related to this further testing, screening, and/or procedures will depend on insurance coverage and there may be some additional costs to you. If you do not have insurance, we will direct you to resources that can help you get insurance for him/her.

You will also be compensated for your time participating in this study. If you agree to take part in this research study, we will pay you/your child a total of $80 in gift cards over the course of your participation for your time and effort. For Study Visit 1, which includes completion of the baseline survey, genetic counseling, and saliva, cheek swab or blood sample collection for your child and his/her parent(s) (if available), we will give you a $20 gift card. For Study Visit 2, which includes the return of your child's genetic test results with a genetic counselor and completion of the return of results survey, we will give you a $20 gift card. For Study Visit 3, which includes completion of the final survey, we will give you a $20 gift card.

Tax law may require the Mount Sinai Finance Department to report the amount of payment you receive from Mount Sinai to the Internal Revenue Service (IRS) or other agencies, as applicable. Generally, this reporting would take place if you receive payments that equal $600 or more from Mount Sinai in a calendar year. You would be responsible for the payment of any tax that may be due.

You should also know that it is possible that products may someday be developed with the help of your child's specimens and data, and there are no plans to share any profits from such products with you, regardless of whether your identifiable information is removed.

POSSIBLE BENEFITS:

It is important to know that you or your child may not get any benefit from taking part in this research. Others may not benefit either. However, possible benefits may be learning about you or your child's secondary findings, such as identifying future conditions that can be treated by your physician. Understanding genetic diversity can help all people benefit from genomic medicine. Helping us learn how we can best communicate information about WGS may help individuals who might choose to have WGS in the future.

REASONABLY FORESEEABLE RISKS AND DISCOMFORTS:

There are risks, discomforts, and inconveniences associated with any research study. These deserve careful thought. In addition to what is described below, there may be unforeseeable risks that occur as a result of genome sequencing and its clinical interpretation.

- **Risks related to answering questionnaires:** You may feel uncomfortable answering questions about your knowledge and understanding of genetic testing. You can choose not to answer questions that make you feel uncomfortable.
- **Risks related to randomization:** We cannot think of any risks specifically related to participation in either study arm. This is important as this randomization will take place prior to you consenting to the study.
- **Risks of loss of private information:** this risk always exists, but there are procedures in place to minimize the risk.
- **Risks of a blood draw:** include pain, bruising, and the slight possibility of infection at the place where the needle goes in. Some people feel dizzy or may faint during or after a blood draw.
- **Risks related to learning genetic information:** There is a chance that you may learn that your child carries a genetic change that may increase the risk for a specific medical condition. If that is the case, we may suggest that other members of the family get tested for the same genetic change, and you may learn that a family member is at risk to develop certain medical conditions or diseases. This knowledge might be upsetting and may cause you to have anxiety or psychological distress. As described above, some of these conditions may have treatment or screening options available, while others may not. You will be asked to think about if you want this information long before the data is available. However, even if you decide you would like this information, it can be upsetting. You may also learn that your child's ancestry or parentage is different than you thought. This may also cause some psychological distress. If your child is found to carry a pathogenic variant in a gene, this may affect your child's reproductive decisions. You will have the opportunity to discuss this with the study's genetic counselor, and will be offered additional genetic counseling resources for your future use.
- **Risks associated with genomic testing:** These tests may not generate accurate results in instances that cannot be predicted. Such instances include but are not limited to: incomplete medical and/or family history, unavailability of critical family members for help with interpretation, inaccurate reporting of family relationships, or technical problems. The results of this test may have significant medical, psychological, and social implications for you and your family. You and your family members may experience anxiety before, during, and after testing.
- **Risks related to privacy:** Your child's privacy is very important to us, and we will use many safety measures to protect it. However, in spite of all of these protections, there is the possibility that the genome sequence data derived may, even when presented without other identifying factors, allow your child to be re-identified. Because your genetic information and

your child's genetic information is unique to you and your child, there is a small chance that someone could trace it back to you or your child. Therefore, this research study cannot promise anonymity, particularly if you choose to publish or share your genome sequence data. The risk of this happening is very small, but may grow in the future. If there is a break in security with the dbGaP database, it may also pose a potential risk to blood relatives. For example, it could be used to make it harder for your child (or a relative) to get or keep a job or insurance. If you or your child's private information was misused it is possible you or your child would also experience other harms, such as stress, anxiety, stigmatization, or embarrassment from revealing information about your or his/her family relationships, ethnic heritage, or health conditions. Specific illnesses and known genetic problems may be found by examining DNA. In the future, insurance companies may use this information to determine if someone is able to be insured by their company. The genetic results from this study will become part of your child's medical record. Insurance companies routinely have access to such records.

- **Group Risks:** Although we will not give researchers your child's name, we will give them basic information such as your race, ethnic group, and sex. This information helps researchers learn whether the factors that lead to health problems are the same in different groups of people. It is possible that such findings could one day help people of the same race, ethnic group, or sex as your child. However, they could also be used to support harmful stereotypes or even promote discrimination.
- **Risks related to insurance:** There is a Federal law called the Genetic Information Nondiscrimination Act (GINA). In general, this law makes it illegal for health insurance companies, group health plans, and most employers of over 15 people to discriminate against your child based on your genetic information. However, it does not protect you or your child against discrimination by companies that sell life insurance, disability insurance, or long-term care insurance.

OTHER POSSIBLE OPTIONS TO CONSIDER:

You may decide not to take part in this research study without any penalty. The choice is totally up to you.

IN CASE OF INJURY DURING THIS RESEARCH STUDY:

If you believe that you have suffered an injury related to this research as a participant in this study, you should contact the Principal Investigator.

ENDING PARTICIPATION IN THE RESEARCH STUDY:

You may stop taking part in this research study at any time without any penalty. This will not affect your ability to receive medical care at any of the Mount Sinai Health System hospitals or elsewhere, or to receive any benefits to which you are otherwise entitled.

If you decide to stop being in the research study, please contact the Principal Investigator or the research staff if you decide you don't want your samples and/or data to be used for research anymore, you can contact the researcher and ask to have your samples and/or data removed from future use. You must do so in writing to the Principal Investigator at the address on the first page. If any samples or data have already been shared without your identity, it won't be possible to retrieve them because no one will know who you are. Samples and data that have already been used will not be affected by your decision. Any samples and/or data that are still linked to your identity by a code the researcher has will be withdrawn so that no future sharing of your samples and/or data will take place. If your samples have already been deposited in an external repository, the study team will request that your samples be removed.

Even if you withdraw your authorization, the Principal Investigator for the research study may still use the information that was already collected if that information is necessary to complete the research study. Your health information may still be used or shared after you withdraw your authorization if you have an adverse event (a bad effect) from participating in the research study.

Withdrawal without your consent: The study doctor, the sponsor or the institution may stop your involvement in this research study at any time without your consent. This may be because the research study is being stopped, the instructions of the study team have not been followed, the investigator believes it is in your best interest, or for any other reason. If specimens or data have been stored as part of the research study, they too can be destroyed without your consent.

CONTACT INFORMATION:

If you have any questions, concerns, or complaints at any time about this research, or you think the research has harmed you, please contact the office of the research team and/or the Principal Investigator at phone number 212-241-8288.

This research has been reviewed and approved by an Institutional Review Board. You may reach a representative of the Program for Protection of Human Subjects at the Icahn School of Medicine at Mount Sinai at telephone number (212) 824-8200 during standard work hours for any of the reasons listed below. This office will direct your call to the right person within the Mount Sinai Health System:

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

DISCLOSURE OF FINANCIAL INTERESTS:

Sometimes, physicians/researchers receive payments for consulting or similar work performed for industry. Effective September 2014 Mount Sinai reviews only payments to an individual totaling more than $5,000 a year per entity when determining potential conflicts of interest. If you have questions regarding industry relationships, we encourage you to talk your physician/researcher or visit our website at http://icahn.mssm.edu/ where Mount Sinai publicly discloses the industry relationships of our faculty.

MAINTAINING CONFIDENTIALITY – HIPAA AUTHORIZATION:

As you take part in this research project it will be necessary for the research team and others to use and share some of your private protected health information. Consistent with the federal Health Insurance Portability and Accountability Act (HIPAA), we are asking your permission to receive, use and share that information.

What protected health information is collected and used in this study, and might also be shared with others?
As part of this research project, the research team at the hospital(s) involved in the research will collect your name and telephone numbers, date of birth, and medical record number.

The researchers will also get information from your child's medical record.

During the study the researchers will gather information by:

- taking a medical history (includes current and past medications or therapies, illnesses, conditions or symptoms, family medical history, allergies, etc.)
- completing the tests, procedures, questionnaires and interviews explained in the description section of this consent.
- reviewing genetic tests

Why is your protected health information being used?
Your personal contact information is important to be able to contact you during the study. Your child's health information and the results of any tests and procedures being collected as part of this research study will be used for the purpose of this study as explained earlier in this consent form. The results of this study could be published or presented at scientific meetings, lectures, or other events, but would not include any information that would let others know who you are, unless you give separate permission to do so.

The research team and other authorized members of The Mount Sinai Health System ("Mount Sinai") workforce may use and share your information to ensure that the research meets legal, institutional or accreditation requirements. For example, the School's Program for the Protection of Human Subjects is responsible for overseeing research on human subjects, and may need to see your information. If

you receive any payments for taking part in this study, the Mount Sinai Finance Department may need your name, address, social security number, payment amount, and related information for tax reporting purposes. I*f the research team uncovers abuse, neglect, or reportable diseases, this information may be disclosed to appropriate authorities.*

Who, outside Mount Sinai, might receive your protected health information?
As part of the study, the Principal Investigator, study team and others in the Mount Sinai workforce may disclose your protected health information, including the results of the research study tests and procedures, to the following people or organizations: (It is possible that there may be changes to the list during this research study; you may request an up-to-date list at any time by contacting the Principal Investigator.)

- Other collaborating research center(s) and their associated research/clinical staff who are working with the investigators on this project: The National Institutes of Health, the Clinical Sequencing Evidence-Generating Research Consortium, and Albert Einstein College of Medicine/Montefiore Medical Center
- Research data coordinating office and/or their representative(s) who will be responsible for collecting results and findings from all the centers: the Clinical Sequencing Evidence-Generating Research Consortium
- Outside laboratory who will be performing laboratory analysis for all the research centers involved in this project: The New York Genome Center
- The sponsoring government agency and/or their representative who need to confirm the accuracy of the results submitted to the government or the use of government funds: National Human Genome Research Institute (NHGRI) and the National Institutes of Health (NIH).
- The United States Department of Health and Human Services and the Office of Human Research Protection.

In almost all disclosures outside of Mount Sinai, your child will not be identified by name, social security number, address, telephone number, or any other direct personal identifier. Some records and information disclosed may be identified with a unique code number. The Principal Investigator will ensure that the key to the code will be kept in a locked file, or will be securely stored electronically. The code will not be used to link the information back to you without your permission, unless the Institutional Review Board allows it after determining that there would be minimal risk to your privacy. The Certificate of Confidentiality obtained from the Department of Health and Human Services will not be used to prevent disclosure to local authorities of child abuse and neglect, or harm to self or others. It is possible that a sponsor or their representatives, a data coordinating office, or a contract research organization, will come to inspect your records. Even if those records are identifiable when inspected, the information leaving the institution will be stripped of direct identifiers. Additionally, *the Office of Human Subjects Protection (OHRP) of the Department of Health and Human Services as well as the Food and Drug Administration (FDA) will be granted direct access to your medical records for verification of the research procedures and data. They are authorized to remove information with identifiers if necessary to complete their task. By signing this document you are authorizing this access.* We may publish the results of this research. However, we will keep your name and other identifying information confidential.

For how long will Mount Sinai be able to use or disclose your protected health information?
Your authorization for use of your protected health information for this specific study does not expire.

Will you be able to access your records?
During your participation in this study, you will have access to your medical record and any study information that is part of that record. The investigator is not required to release to you research information that is not part of your medical record.

Do you need to give us permission to obtain, use or share your health information?
NO! If you decide not to let us obtain, use or share your health information you should not sign this form, and you will not be allowed to volunteer in the research study. If you do not sign, it will not affect your treatment, payment or enrollment in any health plans or affect your eligibility for benefits.

Can you change your mind?
You may withdraw your permission for the use and disclosure of any of your protected information for research, but you must do so in writing to the Principal Investigator at the address on the first page. Even if you withdraw your permission, the Principal Investigator for the research study may still use your protected information that was already collected if that information is necessary to complete the study. Your health information may still be used or shared after you withdraw your authorization if you should have an adverse event (a bad effect) from being in the study. If you withdraw your permission to use your protected health information for research that means you will also be withdrawn from the research study, but standard medical care and any other benefits to which you are entitled will not be affected. You can also tell us you want to withdraw from the research study at any time without canceling the Authorization to use your data.

If you have not already received it, you will also be given The Hospital's Notice of Privacy Practices that contains more information about how The Hospital uses and discloses your protected health information.

It is important for you to understand that once information is disclosed to others outside Mount Sinai, the information may be re-disclosed and will no longer be covered by the federal privacy protection regulations. However, even if your information will no longer be protected by federal regulations, where possible, Mount Sinai has entered into agreements with those who will receive your information to continue to protect your confidentiality.

If as part of this research project your medical records are being reviewed, or a medical history is being taken, it is possible that HIV-related information may be revealed to the researchers. If that is the case, the following information concerns you. If this research does not involve any review of medical records or questions about your medical history or conditions, then the following section may be ignored.

Notice Concerning HIV-Related Information

If you are authorizing the release of HIV-related information, you should be aware that the recipient(s) is (are) prohibited from re-disclosing any HIV-related information without your authorization unless permitted to do so under federal or state law. You also have a right to request a list of people who may receive or use your HIV-related information without authorization. If you experience discrimination because of the release or disclosure of HIV-related information, you may contact the New York State Division of Human Rights at (888) 392-3644 or the New York City Commission on Human Rights at (212) 306-5070. These agencies are responsible for protecting your rights.

Certificate of Confidentiality:
To further protect your privacy, the researchers have obtained a Certificate of Confidentiality from the Department of Health and Human Services. This is intended to ensure that your identity as a participant in this research study will not have to be disclosed as a result from a subpoena, for the purpose of identifying you in any federal, state, or local civil, criminal, administrative, legislative, or other proceedings other than to the FDA or OHRP as identified above.

The research staff will not share any of your research information or biospecimens with anyone who is not a member of the research team, including any family members or friends, other than to those identified above. However, you should know that if we learn that you or someone else is threatened with serious harm, such as a child or an elderly person being abused, the investigators may notify the appropriate authorities if necessary to protect you or others. 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. This means that you and your family must also actively protect your own privacy. If an insurer or employer learns about your research participation, and you agree that they can have your research information, then the researchers may not use the Certificate of Confidentiality to keep this information from them.

ADULT PARTICIPANT:
Your signature below documents your permission to take part in this research and to the use and disclosure of your protected health information. A signed and dated copy will be given to you.

_____	_____	_____	_____
Signature of parent/guardian	Printed Name of parent/guardian	Date	Time

❑ Parent
❑ Guardian (May provide permission only if legally authorized to consent to the child's general medical care.)

_____	_____	_____	_____
Signature of second parent/guardian	Printed Name of second parent/guardian	Date	Time

----------FOR IRB USE ONLY----------
Rev 1.16.19

Note on Second Parent: *If the IRB determined both parents must give permission unless an exception below applies, and if documented permission of the second parent of this child is not obtained, indicate the reason: (select one)*

- ❑ *Second parent is deceased*
- ❑ *Second parent is unknown*
- ❑ *Second parent is incompetent*
- ❑ *Second parent is not reasonably available*
- ❑ *Only one parent has legal responsibility for the care and custody of the child*

PERSON EXPLAINING STUDY AND OBTAINING CONSENT:

_____	_____	_____	_____
Signature of consent delegate	Printed Name of consent delegate	Date	Time

WITNESS SECTION:

When a witness is required to observe the consent process, it should be documented below (for example, when subject is illiterate, visually impaired, or this document accompanies a short form consent).

My signature below documents that the information in the consent document and any other written information was accurately explained to, and apparently understood by, the subject, and that consent was freely given by the subject.

_____	_____	_____	_____
Signature of Witness	Printed Name of Witness	Date	Time