Form Version Date: 11/18/2020

Baseline Survey (Parents of All Participants)

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 you or others. Participation is entirely voluntary. It is completely up to you whether or not you take part. You can also change your mind at any time and it will not affect your ability to get medical care within the Mount Sinai Health System or elsewhere.

The purpose of this research study is learn how genomic testing can help children and young adults with rare diseases. Genomic testing is a way for scientists to study your DNA (genetic material inherited from your parents that at least in part determines your features like eye color, height, and risk of many diseases). Another major goal of this study is to learn the best way to communicate these complicated genomic results back to families likes yours, by having parents answer a series of surveys. Everyone in the study must have a least one parent available to answer these surveys. ***This consent form is focused only on the initial survey.***

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.

The consent form for the entire study will be reviewed with you at your child's first study visit. Signing this initial survey consent does not obligate you to participate in the rest of the study.

If you choose to participate, you will be asked to take part in one study survey, lasting about 45 minutes to one hour. Taking part in this study will not involve added costs to you.

The main risks to you if you choose to allow your child to participate are risks related to the possibility of a loss of confidentiality or privacy. 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.

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 the information you provide may contribute to greater understanding of children like your own, whose conditions are difficult to diagnose and treat. Your answers will help us learn how we can best communicate information about genomic testing to parents and caregivers when their children undergo similar 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 have undiagnosed, likely genetic* cause of neurologic, immunologic, or cardiac disorder(s). If your 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 is expected to last about an hour, or as it takes for you to review the study details and this consent form with the study coordinator. If you decide to participate in the survey studies, and if you decide to participate in the whole study, both you and your child's direct participation will involve

three study visits over the first nine months of this three-year study, and each visit will last about 1 to 2 hours.

The number of people expected to take part in this research study (baseline survey) at Mount Sinai Health System is approximately 250 parents, with another 250 parents participating at the Montefiore Medical Center for a total of 500 parent participants.

DESCRIPTION OF WHAT'S INVOLVED:

If you agree to participate in this research study, the following information describes what may be involved.

Survey
You will be asked to complete an hour-long survey with the study coordinator via a telehealth platform (Zoom), which allows for the completion of this study visit in your own home. This survey will ask you about your past experiences with genetic testing, your understanding and expectations for participating in this study, and some background questions about you and your child. We will also ask some questions about your experiences with the healthcare systems and the level of support and care your child needs. The Zoom link information was emailed to you beforehand along with instructions on how to set-up and use Zoom.

Random assignment

Each participant will have a study visit with a genetic counselor using telehealth. The telehealth platform used in this study provides video and audio support, which will allow you and the study personnel to see and hear each other. Half of the study participants will *also* use a screen-sharing capability on the telehealth while the other half will not. This allows the genetic counselor to present images to you on the screen. The study treatment you will get was chosen by chance, like flipping a coin. You had an equal chance of being given each study treatment. 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.

Scheduling first visit

You will then be given an appointment to have your first study visit with the assigned study genetic counselor, at which time you will review the informed consent for the rest of the study and receive genetic counseling. 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 draw 1-3 Tablespoons of blood from your child. We would also like to take about 3 tablespoons of blood from each biological parent (if available).

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.

COVID-19 procedures related to in-person activities

For in-person activities related to the study, 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:

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.

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:

- Reviewing and signing this informed consent document.
- Answering questions in the initial survey
- Completing an initial genetic counseling visit, which includes providing a blood, saliva or cheek swab sample.

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 permit your child 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 $40 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.

POSSIBLE BENEFITS:

It is important to know that you and your child may not get any benefit from taking part in this research. Others may not benefit either. However, possible benefits may be the information you provide may contribute to greater understanding of children like your own, whose conditions are difficult to diagnose and treat. Your answers will help us learn how we can best communicate

information about genomic testing to parents and caregivers when their children undergo similar testing.

REASONABLY FORESEEABLE RISKS AND DISCOMFORTS:

- **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.
- **Risk 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 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 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 or your child has suffered an injury related to this research as a participant in this study, you should contact the Principal Investigator, Dr. Eimear Kenny.

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 you or your child's ability to receive medical care at any of the Mount Sinai Health System hospitals or elsewhere, or to receive any benefits to which you, or your child, are otherwise entitled.

If you decide to stop your child from being in the research study, please contact the Principal Investigator or the research staff. You may also withdraw your permission for the use and disclosure of any of your child's 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 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 child's health information may still be used or shared after you withdraw your authorization if your child has an adverse event (a bad effect) from participating in the research study.

Withdrawal without your permission: The study doctor, the sponsor or the institution may stop your child's involvement in this research study at any time without your permission. 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 child's 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, address, telephone numbers, email address and date of birth and your child's name, address, telephone numbers, date of birth, and medical record number. The researchers will also get information from your child's Mount Sinai medical record.

During the study the researchers will gather information by:

- completing an initial survey with you
- reviewing your child's electronic medical record to retrieve information about your diagnosis, date of diagnosis, date of genetic test(s), type of genetic test(s), results of genetic test(s), when and where results were delivered to you and by whom, who else was in the room when results were returned and notes of the genetic counselor about the session.

Why is your and your child's 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 and your child's 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, you or 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 your child without your permission, unless the Institutional Review Board allows it after determining that there would be minimal risk to your child's 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 child's 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 child's medical records for verification of the research procedures and data. They are authorized to remove information with identifiers if necessary to complete their tasks. By signing this document you are authorizing this access.* We may publish the results of this research. However, we will keep your child's name and other identifying information confidential.

For how long will Mount Sinai be able to use or disclose your and your child's 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 child's records?
During your participation in this study, you will have access to your child's 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 child's medical record.

Do you need to give us permission to obtain, use or share your or your child's health information?
NO! If you decide not to let us obtain, use or share your/your child's 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/your child's treatment, payment or enrollment in any health plans or affect his/her or your eligibility for benefits.

Can you change your mind?
You may withdraw your permission for the use and disclosure of any of your or your child's 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 child's 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 child's 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.

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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 subject	Printed Name of Subject	Date	Time

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