

Research Consent /Authorization Form

IRB #2011-0320

Study Name: Simons Searchlight

Full Title: Simons Searchlight

Lead Researchers: Cora M Taylor, PhD (Geisinger) and Wendy K Chung, MD, PhD (Columbia University)

Site(s): Geisinger and Columbia University

Study Phone Number: 1-855-329-5638 (Toll-Free)

Funded by: Simons Foundation

We are asking you to be in a health research study.

You are being asked to take part in this research study because you or someone in your family has a genetic change associated with autism spectrum disorders. You do not have to be in this study. Your access to care will not change if you say no. If you join this study, you can stop at any time. This form tells you about the study and how your health information will be used.

What should I do?

- Read this form or have it read to you.
- Contact the study coordinators by email or phone with any questions.
- Take time to think about this and talk to your family and friends.

A description of this study is be available on www.clinicaltrials.gov, as required by U.S. law. This website will not include information that can identify you. At most, the website will include a summary of the results. You can search this website at any time.

During the study, we will tell you if there is new information or changes to the study that could affect you, your health or your desire to stay in the study.

Why is this study being done?

The purpose of this study is to recruit individuals and families with specific genetic changes of interest. These genetic changes may be associated with autism spectrum disorders (ASD) and other developmental disorders. For this study we are collecting information about your development, behavior, and medical history.

Who will be in the study?

About 5000 people will take part in this research study online.

How long will I be in the study?

You will be in the study indefinitely. There is no expiration date for the use and disclosure of your protected health information. You can stop participating in this study at anytime.

When a participant turns 18 years old, he/she will be able to participate in the Simons Searchlight study as an adult. They will need to sign their own consent or be consented by their legally appointed representative (LAR).

What will I be asked to do?

If you take part in this research study, you may be asked to:

- Create an account online at simonssearchlight.org
- Complete consent for yourself and any children that are participating
- Send a copy of all genetic testing results to our team
 - We can help you to get copies of genetic testing results if you do not have them yourself
- Complete surveys online and by phone each year that you participate

- Complete new online surveys as they are added to the study
- Provide biological sample(s) to the Simons Searchlight repository (optional)

What will happen to my biological sample(s)?

You may give biological sample(s) for future research to learn about, prevent or treat health problems. You should not expect to get any results from research done with your sample(s). Your samples will be labeled with a study ID and will not be linked to personal identifying information. Your samples may be accessed by Simons Searchlight staff, Simons Foundation and other approved researchers. This is an optional part of the study.

You may be asked to provide blood, skin cells (biopsy), saliva and/or cheek cells. Your sample(s) will be de-identified and stored at:

Infinity BiologiX (IBX)
145 Bevier Rd.
Piscataway Township, NJ 08854

Your samples may be processed or held at other Simons Foundation-affiliated laboratories before being stored at IBX.

We will keep your sample(s) as long as it is useful. If you allow your sample(s) to be kept for research, you can change your mind at any time. Your sample(s) will be destroyed unless they can no longer be traced to you.

You will not be offered payment for agreeing to have your sample(s) stored. There will be no costs to you for having your sample(s) stored. You will not have to pay to have your sample(s) collected.

The researchers in this study and other individuals who may have access to your sample(s) are not authorized to ever use this material to attempt to clone a human.

The Genetic Information Nondiscrimination Act of 2008 (GINA), makes it illegal for health insurance companies, group health plans, and most

employers to discriminate against you based on your genetic information. This protection does not apply to life insurance, disability insurance, or long-term care insurance.

Biological sample collection:

- **Blood draw:** Four tubes (40mL or less than 3 tablespoons) of blood may be collected during the blood draw. If you agree, blood may be used for research that could include creating cells that can grow indefinitely (cell line).
- **Skin biopsy:** A skin sample no larger than 3mm across may be collected during a skin biopsy. If you agree, skin samples may be used for research that could include creating cells that can grow indefinitely (cell line).
- **Saliva/Cheek cells:** You maybe you may be asked to provide a saliva sample by spitting into a tube. You may be asked to use a buccal swab (a cotton-tipped stick, like a Q-tip) to gently collect cheek cells from the inside of your mouth.

What are the costs?

There will be no costs to you for participating in this study. You will not be charged for any of the research activities.

If the surveys or sample collection lead to injury or outside medical care, you or your insurance company will be charged or held responsible for these costs.

Will I be paid?

Online surveys may be assigned based on age, genetic diagnosis, primary spoken language, or relationship in the family. You will receive a gift card for each online survey completed, ranging from \$5-10 per survey based on complexity of surveys. You will receive \$25-\$50 gift cards for longer surveys completed by phone.

You can stop filling out surveys at any time if you feel uncomfortable. You will need to submit the information you are comfortable providing to receive your gift card.

Can being in this study help me?

This study might or might not help you. We hope that what is learned from this study will help others with rare genetic disorders in the future.

You may learn some information about your child and your family as part of this study. You may receive genetic testing results. You may receive summary information about all participants, or participants with your genetic change.

What are the risks?

There are no physical risks expected with this study. There is a risk that your information could be seen by someone other than the study staff. However, we will take steps to protect your information.

You may feel stress related to sensitive questions. You can stop and talk to study staff or stop answering questions at any time.

You or your insurance will be charged for the diagnosis and treatment of any injury that results from your routine care.

If you have a blood draw or skin biopsy, it may cause pain, bruising, lightheadedness, and on rare occasions, infection. Care will be taken to avoid all complications.

If you have genetic testing as part of this study, it may reveal a genetic change. You will be told if a change is found by a genetic counselor. Genetic counselors help people and families understand genetic testing results and can give you support.

If we do find any genetic information of use to you, we will let you know and may refer your family for further clinical care. You can choose if you want to be contacted if we find meaningful genetic information.

- | |
|---|
| <p><input type="checkbox"/> I would like to be contacted to discuss medically significant findings.</p> <p><input type="checkbox"/> I would NOT like to be contacted to discuss medically significant findings.</p> |
|---|

How will Simons Searchlight and Geisinger use and share my information?

The Simons Searchlight and Geisinger study staff will collect information about you during this study. Some of information will be kept in a research record at Geisinger. Your de-identified records will be kept indefinitely.

Researchers at multiple locations work on this research study. Your information, including personal health information, may be shared across these locations. The locations include:

- Simons Foundation (Data Coordinating Center)
- Geisinger
- Columbia University

Your information, including personal health information, may be shared across other Simons Searchlight sites that were involve in either phase of this study.

Your personal information may be shared with external research services to send information like study newsletters and surveys. This information may include your name and email address.

Your data is stored with a unique study ID with no personal, identifying information. It will be secured and sent to a study database. Only approved study staff can link your data to your personal information.

How will others use and share my information?

The information shared with Simons Foundation and its partners will include:

- Date of Birth
- Study ID number
- Medical history that you have provided
- Information related to you or your family member's genetic diagnosis

The information sent to the Simons Foundation and its partners may be kept and used indefinitely.

Your research and medical record could be reviewed for quality and to make sure rules are followed. This review could be done by:

- Geisinger Institutional Review Board
- Department of Health and Human Services (DHHS)
- Office for Human Research Protections (OHRP)
- Simons Foundation and its partners
- Government agencies in other countries

Your clinical, demographic, and genetic data may be shared with other Simons Foundation studies. These studies include, but are not limited to SPARK, SSC, and other approved studies. This information will be shared using your linked research ID number and using a secure transfer system.

If you agree to participate, you will also be allowing Simons Searchlight to provide de-identified research data to:

- NDAR (National Database for Autism Research)
- Other public databases, which could include but are not limited to ClinVar, dbGaP, or other de-identified data repositories.

If information from this research study is included in an article published in a medical journal or presented at a medical or scientific meeting, it will be done in a way that does not identify you.

How is my information protected?

We will take steps to protect your information. Your data will be stored in a locked file cabinet or password protected computer. We cannot guarantee absolute confidentiality. Your personal information may be disclosed if required by law.

To help protect you and/or your child's privacy, the researchers on this study have a Certificate of Confidentiality from the National Institutes of Health (NIH). The NIH is part of the U.S. Government. With this Certificate, we cannot be forced to share information that may identify you in any US federal, state, or local civil, criminal, administrative, legislative,

or other proceedings. However, the disclosure of you and/or your child's identity may be found necessary, upon request of DHHS, for audit or evaluation.

Information from the research study will not be used to target you for marketing or sales communications.

What if I have questions or problems?

For questions about the research study, contact the study team.

Contact the study team, PI Cora Taylor, or PI Wendy Chung at 1-855-329-5638 (Toll-free) or at coordinator@simonssearchlight.org if you:

- Have questions, concerns or complaints about the study
- Feel you have had a study-related injury

Call the Geisinger Institutional Review Board (IRB) at 1-844-542-3299 or 1-570-271-8663 (Danville, PA)

- If you have questions about your rights as a research participant.
- If you have questions, concerns or complaints about the research.

Consent Signature Page

I agree to take part in this research study and allow my health information to be used for this research study. My questions have been answered. I will be able to review, download and/or print a complete copy of this consent form upon completion.

Do you consent to participate in Simons Searchlight research?

- ☐ Yes
- ☐ No

Optional Biological Sample Collection and Storage:

Please select one of the options below:

- ☐ I voluntarily agree to allow my sample(s) to be stored for use in future research, including DNA tests, to learn about, prevent, or treat health problems.
- ☐ I do not agree to allow my sample(s) to be stored for use in future research, including DNA tests, to learn about, prevent, or treat health problems.

Induced pluripotent stem cells (iPS) cells are a kind of cell that can be made into different cell types from blood or skin. Please select one of the options below related to iPS cells. You can give, store, and test sample(s) without agreeing to IPS cells.

- ☐ I give my permission to have induced pluripotent stem cells (iPS) generated from my donated sample(s).
- ☐ I do not give permission to have induced pluripotent stem cells (iPS) generated from my donated sample(s).

Research Participant's Name
(Auto-filled from participant profile)

Date
(Auto-filled when completed)

CONSENT FOR CHILDREN

Child's Name (Auto-populated from participant profile)

Do you consent for this child to participate in Simons Searchlight research?

☐ Yes

☐ No

If your child is between the ages of 10 and 17, please explain the study to him/her and ask permission to participate in the study – Please select one of the following:

☐ **Not applicable; this child is under age 10.**

☐ **I explained the study to my child. My child agrees to participate in this study.**

☐ **In my opinion, this child is not capable of agreeing to participate in this study.**

Optional Biospecimen Sample(s) Collection and Storage:

Please select one of the options below:

☐ I voluntarily agree to allow my child's sample(s) to be stored for use in future research, including DNA tests, to learn about, prevent, or treat health problems.

☐ I do not agree to allow my child's sample(s) to be stored for use in future research, including DNA tests, to learn about, prevent, or treat health problems.

Induced pluripotent stem cells (iPS) cells are a kind of cell that can be made into different cell types from blood or skin. Please select one of the options below related to iPS cells. You can give, store, and test sample(s) without agreeing to IPS cells.

☐ I give my permission to have induced pluripotent stem cells (iPS) generated from my child's donated sample(s).

☐ I do not give permission to have induced pluripotent stem cells (iPS) generated from my child's donated sample(s).

Parent's/Legal Guardian's Name
(Auto-filled from participant profile)

Today's Date [MM-DD-YYYY]
(Auto-filled when completed)