

## Research Consent & HIPAA Authorization Form

### IRB #2023-1257

**Study Name:** Simons Searchlight

**Full Title:** Simons Searchlight

**Lead Researchers:** Cora M Taylor, PhD (Geisinger) and Wendy K Chung, MD, PhD (Boston Children's Hospital)

**Site(s):** Geisinger and Boston Children's Hospital

**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 available on [www.clinicaltrials.gov](http://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?**

Up to 100,000 participants will participate. Participants include people with an eligible gene change and their families.

## **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 any time.

The investigators may withdraw you from participating in this study if your variant is downgraded or your gene is removed from the list of genes being studied.

## **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 [www.simonssearchlight.org](http://www.simonssearchlight.org)
- Complete consent for yourself and any children or legal dependents that are participating.
- Send a copy of all genetic testing results to our team so that our lab team can review them and confirm eligibility for the study.
  - We can help you to get copies of genetic testing results if you do not have them yourself. You will be asked to sign a medical record release form if you would like our team to request them on your behalf.
- Complete surveys online and a phone interview 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 for storage and future research (optional)
- Provide saliva or a cheek swab for genetic testing (optional).
  - This only applies to specific participants, determined by genetic counselors. If you are determined to be eligible for targeted genetic testing, you will sign a separate consent form.

## **What will happen to my data and biological samples?**

Your data and, if applicable, biological samples will be labeled with a study ID. Your samples may be accessed by Simons Searchlight staff, Simons Foundation and other approved researchers. Only Simons Searchlight staff have the ability to link personal identifying information to your data and sample. Approved researchers will not be able to link identifying information to your data and sample. Some researchers may be sponsored by companies who are trying to make new supports and treatments to learn about, prevent or treat health problems. Your data and/or biological sample may be used for commercial profit. There will be no shared commercial profit with participants.

We will keep your data and/or sample as long as it is useful. If you allow your data and/or sample to be kept for research, you can change your mind at any time. If you change your mind, your data and/or sample will be destroyed unless they can no longer be traced to you. Please note that data already distributed to other approved researchers cannot be recalled and samples distributed to approved researchers will not be destroyed, even if you change your mind about this study.

You will not be offered payment for agreeing to have your data and/or sample stored. There will be no costs to you for having your data and/or sample stored.

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.

## **More information about optional biological samples**

Providing a biological sample is an optional part of this study and if you decide to not provide a biological sample, it does not affect your study participation. Your data and biological sample may be used for future research, without additional informed consent, to learn about, prevent or treat health problems. You should not expect to get any results from research done with you sample.

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

Sampled  
30 Knightsbridge Road  
Piscataway, NJ, USA 08854

Your samples may be processed or held at other Simons Foundation-affiliated laboratories before being stored at Sampled. Sampled was formerly known as both Infinity BiologiX (IBX) and RUCDR.

The following are the sample types that could be collected as part of the optional sample collection that you will decide your participation on at the end of this consent form. These samples are not related to targeted genetic testing that Simons Searchlight offers some participants. If you are eligible for targeted genetic testing, you will be contacted and consent for that separately.

- **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).
- **Saliva/Cheek cells:** 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.

You will not have to pay to have your sample collected.

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

## **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.

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

## **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 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 will be given the option to receive results of some surveys. Results may include information

about your/your child/dependent's behavior and/or development. It is possible you may find results of some surveys upsetting. If this occurs, you should call your doctor or mental health provider to discuss your concerns.

You should know the information we return is not a substitute for a clinical evaluation or feedback and we are not able to monitor responses to surveys. If you have concerns about your spouse's, child's, relative's or your own thoughts, behavior or safety, contact your physician and seek a professional clinical evaluation, or help them to do the same. If you ever have concerns that someone is at risk of harming themselves or others, you should call 911 or take them to the nearest emergency room.

If you have a blood draw 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. A genetic counselor will tell you if a change is found. 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 provide resources for your family for further clinical care. You can choose if you want to be contacted if we find meaningful genetic information.

## **How will Simons Searchlight use and share my information?**

The Simons Searchlight study staff will collect information about you during this study. Your data 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
- Boston Children's Hospital
- Tempus Dynamics and/or third-party contractors with whom there is an established contract to protect participant information.

Your information, including personal health information, may be shared across other Simons Searchlight sites that were involved in the current or previous phases 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.

If the same dependent participant is registered and consented for by different people, research participation will be disclosed to all whom have registered the same dependent. This will be done to prevent duplicate data in the Simons Searchlight study.

## 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 approved studies. These studies include, but are not limited to SPARK, SSC, Autism BrainNet and other approved studies. This information will be shared using your linked research ID number and using a secure transfer system. The data collected will be used for research to learn about, prevent, or treat health problems.

You may be contacted to participate in other approved studies as part of the Research Match program. Participation is optional and does not affect participation in Simons Searchlight.

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)
- SFARI Base (Simons Foundation Autism Research Initiative)
- Other public databases, which could include but are not limited to ClinVar, dbGaP, or other de-identified data repositories.

We will share your information, genetic data and/or samples with approved researchers and it may be published in articles in medical journals or presented at medical or scientific meetings. The information shared is labeled with a code (number), but your name or other identifiable information will NOT be shared. Researchers will be able to publish behavioral or medical data that may be labeled with your specific genetic variant.

## **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](mailto: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.

By signing this form, you are giving Geisinger, Boston Children's Hospital, and Simons Foundation permission to use and share your health information. It can be shared indefinitely for purposes of this study and for future research as explained in this form. Your participation in this study is

voluntary. You may decide to no longer participate or leave the study at any time. If you change your mind about participating, tell us in writing. Write to:

Simons Searchlight

Geisinger - ADMI

120 Hamm Drive

Lewisburg, PA, USA 17837

Email: [coordinator@simonssearchlight.org](mailto:coordinator@simonssearchlight.org)

If you leave the study, information already collected will still be used. Both data and biological samples already distributed cannot be recalled. We will only use and share new information if it is needed to protect your safety or follow with the law.

## 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?

- ☐ 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. 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).

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

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Date  
(Auto-filled when completed)