



**A Multidisciplinary Neurofibromatosis  
Research Program at OHSU  
Consent Form**

IRB#:11616

**TITLE:** A Multidisciplinary Neurofibromatosis Research Program at OHSU

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**STUDY COORDINATOR:** Hadley Morotti, MS, (503) 494-2971

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**PURPOSE:**

“You” means you or your child throughout this form.

The purpose of this study is to learn more about people living with neurofibromatosis and related conditions. Neurofibromatosis is one of the most common neurological disorders. Even though risk factors for this disorder are known, early diagnosis and intervention are still challenging. In addition, many individuals with neurofibromatosis may also have additional developmental challenges, such as autism. The goals of our study are to 1) Examine the development and medical history of individuals with neurofibromatosis that do or do not also have autism or related conditions; 2) to find new genes that may be important for people with atypical neurofibromatosis. and; 3) store data and samples in a repository for future studies of neurofibromatosis and related conditions.

Genes are the units of DNA—the chemical structure carrying your genetic information—that determine many human characteristics such as the color of your eyes, your height, and whether you are male or female. Genetic causes likely play a role in many disorders like neurofibromatosis; however, there are likely many different genetic risks for these disorders. We hope to determine how different gene changes might lead to these disorders and identify new subtypes within them.

If the steps that lead from the gene change to symptoms of these disorders can be found,

treatments may be improved. Also, by identifying new subtypes within these disorders, future treatments potentially could become customized to a patient's unique genetic makeup.

You have been invited to participate in this study because we believe that you may have neurofibromatosis or a related condition. We are asking you to provide samples and information for this research. The samples you provide will be processed and stored in a repository. Samples stored in the repository will also be made available for other research studies. Your samples and information will be stored indefinitely. In the future, your samples and information may be given to other researchers as part of the ongoing search for more information on autism. This may include depositing information obtained from your DNA, your medical records, and other assessments in public research databases.

This repository was designed to collect biological samples and information from people with developmental disorders and their relatives. Information will be stored in a registry. The registry is a database that will track medical and family history information and will allow us to contact participants with important new information about these disorders, including research study opportunities.

Through using these samples and information for this study and in future studies, we hope to help identify the underlying causes leading to developmental disorders and to develop new, personalized treatments.

### **WHAT SPECIMENS/DATA WILL BE COLLECTED?**

The specimens collected may include blood, saliva, and discarded or previously stored surgically removed tissue. The study staff will clarify which of the following specimens and information you will be asked to provide.

We will ask for information about your health history, daily life, and family history. We may ask you to complete additional tests related to learning and memory, social interactions, and daily living skills. We may ask your permission to contact your health care provider to obtain information about your health, such as psychological assessments, reports from any clinical assessments, brain scans, and genetic test results. If we need this information, we will ask you to sign a medical record release form giving us permission to obtain this information from your provider.

### **HOW WILL SPECIMENS/DATA BE COLLECTED?:**

Taking part in this study **may** involve the following:

- 1) Blood draw. We may collect a blood sample from a vein in your arm (up to four teaspoons).
- 2) Saliva collection. You will spit into a collection tube or we will swab your mouth. Approximately one teaspoon of saliva is required.
- 3) Tissue collection. If you have tissue removed as part of your medical care, such as during a surgery, we may ask for discarded ("leftover") or previously stored tissue to be transferred to us and added to the repository.
- 4) Medical history information. We will collect health information from your medical record. You may be asked to sign a separate form for the release of medical records.
- 5) Testing. You may be given questionnaires, forms, or asked questions by a researcher about your health and current skills. We may ask you to complete additional tests related to

learning and memory, social interactions, and daily living skills. These may be obtained by phone, mail, or online.

- 6) Information about your relatives. You may be asked to give us health information about your relatives. Any information you give us will be kept strictly confidential. We will not contact your relatives without their permission. We may discuss with you the possibility of including your relatives in the study in the future.

The study visit will take between one and two hours and will involve at most one visit to OHSU. If the consent forms are reviewed with you by phone, then you will need to visit your local doctor or laboratory for sample collection or a saliva collection kit will be mailed to you. We will work with your local doctor or laboratory to ensure there is no cost to you for a blood draw.

### **WHAT WILL HAPPEN TO THE SPECIMENS/DATA?**

The specimens you give will be stored in a secure area in the O'Roak Laboratory at OHSU. Your medical information will be entered in a secure database, and paper copies will be stored in locked file cabinets. A code number will be assigned to your specimens and information. Specimens will be labeled only with this code number. Your stored medical information will include your name, contact information, and other identifiers. Only the investigators listed on this form will be able to link your code number to your name and other personal information and have access to your medical information. Researchers on this study may contact you in the future to update information about you, to inform you about new research opportunities, or to share research results.

In the future, your samples and information may be shared with other researchers at OHSU and elsewhere. This may include depositing information obtained from your DNA, your medical records and other assessments in public research databases, such as, but not limited to, the National Center for Biotechnology Information (NCBI) database of Genotypes and Phenotypes (dbGaP).

In these cases, your samples and information will only be labeled with a code number. Identifying information, such as your name, social security number, or date of birth, will be removed. Other researchers will not be able to link your name to the code number. If scientifically necessary, they may also be given information about the date the samples and information were obtained.

Specimens and information will be stored in this repository indefinitely. If in the future you decide you no longer want to participate in this research, we will remove your name and any other identifiers from your samples and medical information but the material will not be destroyed and we will continue to use it for research.

### **ACCESS TO RESEARCH TEST RESULTS:**

If we discover information that is important for your health care (meaning a potential genetic explanation for your developmental disorder, or genetic information that would require action to treat or prevent a disease), either in this study or in the future, we will contact you and ask whether you wish to receive the results.

If you choose to receive the results, you may be required to have the test repeated in a clinical laboratory. You may learn information about your health that is upsetting or that impacts your family planning, family relationships, insurance, or career. Because genetic information is complex and sensitive, the results should be discussed with a genetic counselor or your primary care provider who can answer your questions or discuss your concerns. You would be responsible for all costs associated with having the test repeated and visiting a doctor or genetic counselor to discuss the results. The costs for the repeat testing and the counseling necessary to be certain

that you understand what the results mean may be billed to you or to your third party carrier. (Caution: this will probably make the results available to the third party carrier and to your clinical record. You may choose to pay out of pocket instead.)

We may contact you again in the future to update your information in the repository.

### **PRIVACY AND CONFIDENTIALITY PROTECTIONS:**

We will take steps to keep your personal information confidential, but we cannot guarantee total privacy. We will create and collect health information about you as described in the above sections of this form. Health information is private and is protected under federal law and Oregon law. By agreeing to be in this repository, you are giving permission (also called authorization) for us to use and disclose your health information as described in this form.

The investigators, study staff, and others at OHSU may use the information we collect and create about you in order to maintain and oversee this study and research repository and to conduct future research projects.

We may release this information to others outside of OHSU who are involved in conducting or overseeing research, including the Office for Human Research Protections (OHRP). OHRP is a federal agency that oversees research involving humans. OHRP is permitted to review and copy your records.

We will not release information about you to others not listed above, unless required or permitted by law. We will not use your name or your identity for publication or publicity purposes, unless we have your special permission.

Under Oregon law, suspected child or elder abuse must be reported to appropriate authorities. OHSU complies with Oregon state requirements for reporting certain diseases and conditions to local health departments.

When we send specimens or information outside of OHSU, they may no longer be protected under federal or Oregon law. In this case, your specimens or information could be used and re-released without your permission.

Data or specimens from this study may be shared with other investigators for future research studies. A code number will be assigned to you or your child's samples and genetic information, as well as to other medical information about you. All data is maintained in a locked area. Only the investigators named on this consent form will be authorized to link the code number to you. Other investigators who may receive samples of your DNA, blood, saliva, tissue, genetic or medical information for research will be given only the code number. If scientifically necessary, they may also be given information about the date the samples and information were obtained. These outside collaborators will sign a data access agreement, agreeing not to attempt to identify you and to help protect your confidentiality.

Your genetic information may be shared in a public online database for future research. The database may contain the date that the samples and information were obtained, but will not contain any other information that directly identifies you, such as your name, address, or birth date, so it is unlikely that someone would know the genetic information came from you. In the future, people may develop ways to identify you or your blood relatives from this information, but currently, there is not a way to identify you without having additional information to compare to it, such as

information from your DNA sample.

We may continue to use and disclose your information as described above indefinitely.

### **RISKS AND DISCOMFORTS:**

Although we have made every effort to protect your identity, there is a small risk of loss of confidentiality. If the results of these studies of your genetic makeup were to be accidentally released, it might be possible that the information we will gather about you as part of this study could become available to an insurer or an employer, or a relative, or someone else outside the study. Even though there are discrimination protections in both Oregon law and federal law, there is still a small chance that you could be harmed if a release occurred.

A federal law, called the Genetic Information Nondiscrimination Act (GINA), generally makes it illegal for health insurance companies, group health plans, and most employers to discriminate against you based on your genetic information. Be aware that this federal law does **not** protect you against genetic discrimination by companies that sell life insurance, disability insurance, or long-term care insurance. GINA also does not protect you against discrimination if you have already been diagnosed with the genetic disease being tested.

#### Risks related to blood draw:

Blood drawing will cause some pain and carries a small risk of bleeding, bruising or infection at the puncture site.

Since “leftover” tissue will only be collected if you are having surgery for another reason, you will be counseled separately about these procedures by the clinician or investigator performing the procedure.

We may ask you questions about you or your child that you may find personal, embarrassing, or upsetting. You may refuse to answer any of the questions that you do not wish to answer.

### **BENEFITS:**

There will not be any direct benefits to you if you decide to participate in this study and research repository. Research conducted on these specimens and data may help researchers to better understand health conditions in the future.

### **COSTS:**

There will be no cost to you or your insurance company to participate in this study.

### **LIABILITY:**

If you believe you have been injured or harmed while participating in this research and require immediate treatment, contact Brian J. O’Roak, PhD at (503) 494-2971.

If you are injured or harmed by the study procedures, you will be treated. Neither OHSU nor the Edna S. Green Trust offers any financial compensation or payment for the cost of treatment if you are injured or harmed as a result of participating in this research. Therefore, any medical treatment you need may be billed to you or your insurance. However, you are not prevented from seeking to collect compensation for injury related to negligence on the part of those involved in the research. Oregon law (Oregon Tort Claims Act (ORS 30.260 through 30.300)) may limit the dollar amount that you may recover from OHSU or its caregivers and researchers for a claim relating to care or research at OHSU, and the time you have to bring a claim.

If you have questions on this subject, please call the OHSU Research Integrity Office at (503) 494-7887.

### **COMMERCIAL DEVELOPMENT:**

By consenting to participate, you authorize the use of your samples and information for the research described in the PURPOSE and PROCEDURES sections of this document. In addition, you acknowledge that OHSU may make any lawful use of your samples and information, including, but not limited to, future research studies, destroying them, or transferring them to a public or private entity.

Samples and information obtained from you in this research may be used to make a discovery that could be patented or licensed to a company. There are no plans to provide financial compensation to you should this occur. However, should OHSU ever provide your samples or information to anyone else for research or commercial use, it will do so in such a way as to protect your privacy and confidentiality as stated in the CONFIDENTIALITY section of this document. Further, you will have no responsibility or liability for any use that may be made of your samples.

### **PARTICIPANT RIGHTS:**

If in the future you decide you no longer want to participate in this study and research repository, we will remove your name and any other identifiers from your biological samples and/or other information, but the material will not be destroyed and we will continue to use it for research.

This research is being overseen by an Institutional Review Board (“IRB”). You may talk to the IRB at (503) 494-7887 or [irb@ohsu.edu](mailto:irb@ohsu.edu) if:

- Your questions, concerns, or complaints are not being answered by the research team.

- You want to talk to someone besides the research team.
- You have questions about your rights as a research subject.
- You want to get more information or provide input about this research.

You may also submit a report to the OHSU Integrity Hotline online at <https://secure.ethicspoint.com/domain/media/en/gui/18915/index.html> or by calling toll-free (877) 733-8313 (anonymous and available 24 hours a day, 7 days a week).

You do not have to join this or any research repository. You do not have to allow the use and disclosure of your health information for this repository, but if you do not, you cannot join the repository or this study.

If you do join the study and repository and later change your mind, you have the right to quit at any time. This includes the right to withdraw your authorization to use and disclose your health information. If you choose not to join this repository, or if you withdraw early from the repository, there will be no penalty or loss of benefits to which you are otherwise entitled, including being able to receive health care services or insurance coverage for services. Talk to the investigator if you want to withdraw from the repository.

If you no longer want your health information to be used and disclosed as described in this form, you must send a written request or email stating that you are revoking your authorization to:

**Brian J. O’Roak, PhD**  
**3181 SW Sam Jackson Park Rd.**  
**Mail Code: L103**  
**Portland, OR 97239-3098**  
**oroak@ohsu.edu**

Your request will be effective as of the date we receive it. However, health information collected before your request is received may continue to be used and disclosed to the extent that we have already acted based on your authorization.

If you have any questions, concerns, or complaints regarding this research repository now or in the future, contact Dr. Brian J. O’Roak at 503-494-2971.

**SIGNATURES:**

Your signature below indicates that you have read this entire form and that you agree to participate in this study and store your specimens and data in this research repository for future research purposes. We will give you a copy of this form.

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| Subject Printed Name | Subject Signature (if 18+ years and<br>No Legally Authorized Representative) | Date |
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| Parent, Guardian, LAR Printed Name | Parent, Guardian, LAR Signature | Date |
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|---------------------------------------|------------------------------------|------|
| Person Obtaining Consent Printed Name | Person Obtaining Consent Signature | Date |
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| Name of Authorized Research<br>Representative | Authorized Research Representative<br>Signature | Date |
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