RESEARCH CONSENT FORM

For Parents/Guardians and Adult Participants
(Online Form)

TITLE: Global A-T Family Data Platform

PROTOCOL NO.: None
WIRB® Protocol #20162677

SPONSOR: Ataxia Telangiectasia Children's Project

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STUDY-RELATED PHONE NUMBER(S): Jennifer Thornton, MSW
954-481-6611
After Hours Voicemail: 954-481-6611

NOTE: If you are a parent or guardian of a minor with ataxia-telangiectasia or a legally authorized representative, the words "you" and "your" below refer to the person in the study rather than to you.

KEY POINTS

"The Global A-T Family Data Platform" initiative is a participant-driven effort, overseen by ataxia-telangiectasia (A-T) research advocates, that will empower people with A-T and their families to help transform research and treatment of the disease by sharing their genetic data and related health information with researchers around the world. If families also agree to allow researchers to contact them with follow-up research questions, the platform will enable the A-T community to have a continuous dialogue with researchers and potentially accelerate progress in A-T research. The ultimate goal of this effort is to create an environment in which the A-T community and their families contribute their clinical and genetic data and govern the sharing of these data with researchers to better understand the disease.

1. What is the purpose of this research initiative?

We (the sponsors) want to better understand genetic and other biological and lifestyle-related factors affecting people with A-T so that we can work toward developing more effective therapies and diagnostics. By partnering directly with people who have A-T, and enabling an ongoing dialogue with researchers, we will be able to study many more cases worldwide and create a comprehensive and evolving resource for A-T genetics and health characteristics. This is a critical step toward developing potential therapies and improving care.

2. What will I have to do if I agree to participate in this effort?

- You will answer some questions about how your disease has affected you and your medical care.

- If you are willing, you will give us permission to contact your doctor to get copies of your medical records, and possibly to confirm your diagnosis.
- If you agree, you will periodically participate in additional research studies and questionnaires about your medical condition and daily life written by researchers who are studying your data. You may choose not to receive these invitations or decline to answer any individual questions.
- If you are willing, you will participate in the genome sequencing study currently open for enrollment, and you will send a saliva sample to us in a pre-stamped package that we will provide.

3. Do I have to participate in this initiative?

No. Taking part in any portion of this initiative is voluntary. If you decline to participate, there will be no penalty to you and you won't lose any benefits. Even if you decide to participate, you can always change your mind and ask that your data no longer be shared. You can also choose not to receive additional communication about future studies. Your medical care will not change whether you participate or not.

4. Will I benefit from participating?

Participating in this study is unlikely to directly or immediately improve your own health. However, the information we collect may aid in research efforts to one day provide better A-T treatment and prevention options. We will provide updates on our website about key research discoveries made possible by your participation. We will not be able to provide you with any specific information about your own genetic data. If, at some point in the future, we believe that analysis of your genetic results may impact or change your diagnosis of A-T, we will contact your doctor, if you have agreed to this beforehand.

5. What are the risks of taking part in this research initiative?

There is a potential risk that your information (which includes information from your health records and possibly your genetic information) could be seen by unauthorized individuals. An accidental release of your genetic information could be used to identify you and your family members. However, we have tried to minimize this risk by carefully limiting access to the computers that would house your information to the staff of this research initiative as well as by creating secured access-controlled software that will hold information about your personal identity (e.g. name, date of birth, address) in an encrypted manner.

Filling out the questionnaires could lead you to feel uncomfortable or upset, in which case please tell the study staff. You have the right to refuse to answer any questions.

6. Will it cost me anything to participate in this research initiative?

No.

7. Who will use my samples and see my information?

After removing your name and other readily identifiable information (this identifying information will be only available to study staff at the Global A-T Family Data Platform who have signed confidentiality agreements and have received special training), your genomic data and health information will be made available to researchers around the world through a secure, access-controlled data repository developed and operated by the Broad Institute of MIT and Harvard. Your data can only be accessed by qualified investigators who have been granted permission by a data access committee selected by the Global A-T Family Data Platform, comprised of A-T family members as well as scientific and medical advisors. We will keep track of every study that has used your information.

Be aware that your study records (which include your medical records, your signed consent form, and other information) will be shared as needed for the study with groups that provide study and regulatory oversight. For example, Western Institutional Review Board® (WIRB®) may look at your study and medical records. WIRB is a group of people who review research studies to protect the rights and welfare of research participants.

8. Can I stop taking part in this research initiative?

Yes, you can withdraw from this research study at any time, and your data will no longer be shared with researchers in future research studies. However, any of your information that has already been shared with investigators cannot be withdrawn from their studies.

9. What if I have questions?

If you have any questions, concerns, or complains please send an email to support@atfamilies.org or call the phone number on the first page of this form, and ask to speak with one of the Global A-T Family Data Platform staff trained on the platform about this initiative and the data platform or if you feel you have experienced a research-related issue.

If you have questions about your rights as a research subject or if you have questions, concerns, or complaints about the research, you may contact:

Western Institutional Review Board® (WIRB®)
1019 39th Avenue SE Suite 120
Puyallup, Washington 98374-2115
Telephone: 1-800-562-4789 or 360-252-2500
E-mail: Help@wirb.com

WIRB is a group of people who perform independent review of research.

WIRB will not be able to answer some study-specific questions, such as questions about appointment times. However, you may contact WIRB if the research staff cannot be reached or if you wish to talk to someone other than the research staff.

THE GLOBAL A-T FAMILY DATA PLATFORM
FULL RESEARCH CONSENT FORM

A. Introduction

You are being invited to participate in a new initiative that will collect and analyze biological samples and health information from people with ataxia-telangiectasia (A-T). The goal of this initiative is to create a data platform that can be accessed by any qualified researcher authorized by an A-T data access board and to enable researchers to have a continuous dialogue with the A-T community. This data may help researchers better understand which genetic and health factors affect A-T and potentially develop ways to treat and prevent it. At least 500 people with A-T are expected to participate in this research initiative.

You are being asked to participate in this initiative because you were diagnosed with A-T. If you join the initiative, we will ask you to answer questions about your medical condition and to share your medical records. Periodically, we will post new questionnaires about your medical condition written by researchers who are studying your data. You may choose not to receive the new questionnaires, and you may decline to answer individual questions.

You will also be invited to participate in a genome sequencing study. The sponsor wants to use some of your saliva to look at your genes or "DNA." DNA is in your cells, and it is what makes each of us unique. DNA controls things like the color of your hair or eyes. DNA might make you more likely to get certain diseases or affect whether a drug helps you and/or gives you side effects.

If you choose to participate in the genomic study, you will submit a saliva (spit) sample and allow us to study your genome using a technology called "DNA sequencing." DNA sequencing is a way of reading the DNA to identify alterations that may contribute to the makeup and behavior of cells. Your genomic data and health information will be studied along with information from other people with A-T in this study, and it will be stored for future studies.

This form describes this research initiative in order to help you decide if you want to participate. This form will tell you what you will have to do if you join, and the risks and benefits of participating.

If you have any questions or do not understand something on this form, you should ask the study staff. You should also discuss your participation with anyone you choose in order to better understand this project and your options. Do not sign this form until your questions have been answered and you decide that you want to be part of this project.

B. What is the purpose of this research initiative?

We want to understand A-T better with the hope that we can develop therapies for this disease and improve the standard of care for people with A-T. By partnering directly with people worldwide, we will be able to create the first comprehensive repository of A-T health and genomic data, a critical step toward research and development of therapies. In addition, we will make this data available in a way that any qualified investigator around the world will be able to conveniently access and analyze it. Moreover, we will enable the researchers to contact participants who agree with follow-up research questions and thereby accelerate their potential to discover new findings about A-T. By doing so, we hope to engage the best minds in the effort to cure A-T.

C. Who is sponsoring this research initiative?

The A-T Children's Project, the sponsor of this study, is paying for this study. The A-T Children's Project is a nonprofit organization that raises funds through the grassroots efforts of friends and families of children with A-T to support and coordinate biomedical research projects, scientific conferences and a clinical center aimed at finding life-improving therapies and a cure for A-T. The principal investigator is an employee of the sponsor. If you have questions or concerns about this, ask the principal investigator for more information.

D. What other options are there?

Taking part in any portion of this research initiative is voluntary – your alternative is not to participate. Your decision not to participate will not affect your medical care in any way or result in any penalty or loss of benefits.

E. What is involved in the research initiative?

To join this initiative, you will need to answer some questions about your health, the doctors you have seen, and your medical and family history. If you choose to participate in the genome sequencing study currently opened for enrollment, you will send a saliva sample to us in a pre-stamped package that we will provide. If you agree, we may obtain copies of your medical

records from hospitals or centers where you received your medical care. We may also provide a way for you to upload your medical records to our platform yourself.

We will analyze the DNA in your cells (obtained from the saliva sample). No additional procedures will be required.

We will link the results of the genetic analysis on your saliva with medical information that has been generated during the course of your treatment as well as with the medical information you will provide in our online questionnaires.

Periodically, we will post questionnaires about your medical condition written by researchers who are studying your data to accelerate progress in understanding A-T. We will also offer you the option to participate in additional future research studies. You may choose not to answer these questionnaires and not to participate in these studies.

We may also share your information with other qualified researchers for use in their own studies involving A-T, studies that have not yet been designed, studies involving diseases other than A-T, and/or studies that may be for commercial purposes (such as the development or approval of new drugs). We may deposit your data in central data repositories genome-wide association studies to enable sharing with the research community. Any future research will be subject to review and oversight requirements as applicable according to federal research regulations. Your information will be stored only with a code number attached. Your name, personal identifying number (such as social security number in the United States, or Health Insurance Number in Canada), and other information that could readily identify you will not be shared with central data banks or other researchers.

Before being able to access your information or request to re-contact you with follow-up questionnaires, researchers will have to apply for permission to a data access committee, comprised of scientific experts and A-T family representatives, who will ensure that the research proposed by the researcher is consistent with the principles laid out in this consent form.

In some cases, if you agree, a research doctor or investigator may contact you to find out if you would be interested in participating in a different or future research study (related to A-T or other medical condition) based on information that may have been found in your saliva sample or other information you provided. You will be given an option at the end of this form to indicate whether or not you wish to be re-contacted.

If the study staff learns any new information that might change your mind about continuing in the study, the study staff will tell you about it.

F. Who is eligible to participate in this initiative?

Any person who has been diagnosed with A-T may participate. We may need to contact your doctor to confirm your diagnosis.

G. How long will I be in this research initiative?

Your direct involvement will be limited initially to the time it takes to obtain your consent and to collect samples and medical information. Because this project involves a data repository, your genetic data and health information will be stored indefinitely for future use. If you agree, we may also re-contact you to get updates about your health and medical care or to tell you about other studies.

Your saliva samples will be stored refrigerated in a secure laboratory in the United States and used for genome sequencing. After your genome sequencing has been completed successfully, your saliva samples will be appropriately discarded.

H. What kind of information could be found in this study and will I be able to see it?
This research is being done to add to our knowledge of A-T and is not intended to provide information about your individual health or treatment. You will not receive the results of the genetic tests. The results will not be added to your regular medical record.

In the unlikely event that the results suggest that your A-T mutations should undergo additional review by your doctor, you may request (at the end of this form) that the study staff contact your doctor and share this information.

If, at some point in the future, these plans change and there is an opportunity to return individual genetic test results and you would like to be informed of your personal results, you can indicate this at the end of this form. In the event of such a change, the study staff would contact you to explain the risks and benefits of obtaining research results and to obtain your separate consent to provide such results to your physician. If you change your mind in the future, you may give your consent to be re-contacted.

We will provide general results and major discoveries to all participants. We will do this by regularly updating the website that you used to learn about this study. Furthermore, we or the researchers we have allowed to access your data will publish important discoveries found through these studies in scientific journals so that the entire research community can work together to better understand A-T. Your individual data will not be published in a way in which you could be readily identified. Abstracts, which are plain language summaries of the published reports, will be available to you and the general public.

I. What are the risks or discomforts that can result from your participation?
There is a small risk that by participating in this research initiative, your genetic and medical information could be seen by unauthorized individuals. An accidental release of your genetic information could be used to identify you and your family members. We have tried to minimize this risk by carefully limiting access to the computers that would house your information to the staff of this research initiative as well as by creating secured access-controlled software that will hold information about your personal identity (e.g. name, date of birth, address) in an encrypted manner.

There are laws to protect privacy and to prohibit the misuse of your information. However, it is important to know that there is still a risk someone could get access to the information we have stored about you, including information about a family member (for example, revealing that you or a blood relative carry a genetic disease).

J. What are the benefits of the research initiative?
Participating in this study is unlikely to directly or immediately improve your own health. However, the information we collect may aid in research efforts to one day provide better A-T treatment and prevention options. Research performed with your genetic and health information could result in the improvement of medical care and the discovery and development of new drugs, tests or commercial products. There are no plans to pay you if tests or products are developed from this research.

K. Can I stop being in the research initiative and what are my rights?

You can stop being part of any portion of this initiative at any time.

If you tell us that you want to stop being in the study, we will destroy any saliva samples or DNA samples we have. We will not perform any additional tests on the samples. Additionally, we will not collect any additional medical records, and we will stop sharing your data with researchers and your data will not be included in any future research studies. We will no longer contact you regarding future research studies. We will not be able to take back the information that already has been used or shared with other researchers, or that was included in analysis that has already been publicly presented.

Participants who enroll in the study when they are under 18 years old will be re-contacted when they turn 18 to reconfirm their consent to continue to use their data in the data repository. At that time, the participant can decide to continue in this project, or ask that his/her data no longer be shared. If such a participant cannot be contacted, the sponsor will continue to use any data already collected.

To withdraw your permission, you must do so in writing by contacting the principal investigator listed on the first page of this form or by updating your profile on the user portal. If you choose to not participate, or if you are not eligible to participate, or if you withdraw from this research study, this will not affect your present or future care and will not cause any penalty or loss of benefits to which you are otherwise entitled.

For adults considering whether to be in the study:

What if I work for the study center or sponsor? What if I am a family member of someone who works for the study center or sponsor?
Study center/sponsor employees and their family members do not have to be in this study. No one should influence or pressure you to be in this study. An employee's or his/her family member's decision to be in the study, or to leave the study early, will not affect the employee's job or job benefits.

For parents/guardians who are considering whether to allow their child to be in the study:

What if I work for the study center or sponsor? What if I am a family member of someone who works for the study center or sponsor?
Study center/sponsor employees and their family members do not have to let their children be in this study. No one should influence or pressure you to let your child be in this study. An employee's or his/her family member's decision to allow a child to be in this study, or to have the child leave the study early, will not affect the employee's job or job benefits.

L. Will I be paid to take part in this research initiative?
You will not be paid to take part in this research initiative.

M. What are the costs?
There are no costs to you to take part in this research initiative.

Your usual health care benefits will not be altered due to your participation in this research initiative.

N. What happens if I am injured or sick because I took part in this research study?

As described above, the risks of being part of this research initiative are mostly related to privacy. However, please contact us if you think you have been injured as a result of participating in any portion of this research.

O. What about confidentiality?

We will take rigorous measures to protect the confidentiality and security of all your information, but we are unable to guarantee complete confidentiality.

If you choose to have us send you a sample collection kit to collect your saliva, your name, personal identifying number (such as social security number in the United States, or Health Insurance Number in Canada), and other information that could be used to readily identify you will be removed and replaced by a code. When your sample is sent to a lab for DNA sequencing, the samples will be identified using only this code.

The medical records that we may receive will be reviewed by the study staff and their medical advisors to obtain information about your medical condition and treatment.

We will store all of your identifiable information related to the study (including your medical records) in locked file cabinets and in password-protected computer files at the A-T Children's Project office and we will limit access to such files. Identifiable information will be logged in a secured access controlled web-based software during enrollment and only the study staff will be able to access it. We may share your identifiable information or coded information, as necessary, with federal regulatory or oversight authorities, and WIRB, which review the conduct of the study, or as otherwise required by law.

When we send the results of the DNA sequencing and your medical information to central data banks or other researchers, they will not contain your name, personal identifying number (such as social security number in the United States, or Health Insurance Number in Canada), or other information that could be used to readily identify you.

The results of future research studies using the information from this data repository may be published in research papers or included in presentations that will become part of the scientific literature. You will not be identified in publications or presentations.

Please note that the entities that process and store your data for this research initiative are located in the United States, and potentially other countries where the laws do not protect your privacy to the same extent as the laws in your country of residence. In such cases, the storage of your data may be subject to the less restrictive data protection laws of these foreign countries rather than the laws of your own country. However, all reasonable steps will be taken to protect your privacy.

P. Whom do I contact if I have questions about any portion of this research initiative?

If you have questions, concerns, or complaint about the initiative, or if you feel you have experienced a research-related issue, please contact the research staff listed on the first page of this form, or by emailing support@atfamilies.org or calling +1 954-481-6611/Toll Free within the United States and Canada: 800-5-HELP-A-T (800-543-5728).

You can ask questions about the study at any time. You can call the study staff at any time if you have any concerns or complaints. You should call the study staff at the phone number listed on page 1 of this form if you have questions about the study procedures.

WIRB reviewed this study. WIRB is a group of people who review research studies to protect the rights and welfare of research participants. Review by WIRB does not mean that the study is without risks. If you have questions about your rights as a research participant, if you are not able to resolve your concerns or complaints with the study staff, if you have a complaint, or if you have general questions about what it means to be in a research study, you can call WIRB or visit the WIRB website at www.wirb.com.

Western Institutional Review Board® (WIRB®)
1019 39th Avenue SE Suite 120
Puyallup, Washington 98374-2115
Telephone: 1-800-562-4789 or 360-252-2500
E-mail: Help@wirb.com

WIRB will not be able to answer some study-specific questions, such as questions about appointment times. However, you may contact WIRB if the research staff cannot be reached or if you wish to talk to someone other than the research staff.

Q. Participation Information

If you decide to sign this consent form, we will ask you for information about contacting your physicians and any hospitals at which you have received treatment for your medical condition. We will not disclose details about the results of your participation in this study with any of the individuals whom we contact, but will ask them to provide us with your medical history.

R. Authorization to use your health information for research purposes

Because information about you and your health is personal and private, it generally cannot be used in any research study without your written authorization. Federal law requires that your health care providers and healthcare institutions (hospitals, clinics, doctor's offices) protect the privacy of information that identifies you and relates to your past, present, and future physical and mental health conditions ("protected health information").

You must authorize this use and sharing of your information by signing this form or you cannot be in the study. You can still be in the main part of the study even if you do not authorize the use and sharing of your information for the optional part(s) of the study.

If you sign this form, it will provide your health care providers and healthcare institutions the authorization to disclose your protected health information to the study researchers for use in this data repository. The form is intended to inform you about how your health information will be used or disclosed in the study. Your information will only be used in accordance with this authorization form and the informed consent form and as required or allowed by law. Please read it carefully before signing it.

1. **What personal information about me will be used or shared with others during this research?**
    a. Your medical records if you choose to share them
    b. Your saliva samples and genome sequence if you choose to participate in the genetic study
    c. New health information created from study-related tests and/or questionnaires

2. **Why will protected information about me be used or shared with others?**
    The main reasons include the following:

a. To conduct and oversee the research described earlier in this form
b. To ensure the research meets legal, institutional, and accreditation requirements
c. To conduct public health activities (including reporting of adverse events or situations where you or others may be at risk of harm)

3. **Who will use or share protected health information about me?**
   The Global A-T Family Data Platform study staff will use and/or share your protected health information in connection with this research study.

4. **With whom outside of the** Global A-T Family Data Platform **may my protected health information be shared?**
   While all reasonable efforts will be made to protect the confidentiality of your protected health information (i.e. identifiable data such as your name and address), it may also be shared with the following entities:
   a. Federal and state/provincial, or similar agencies (for example, the Department of Health and Human Services, the Food and Drug Administration, Health Canada, the National Institutes of Health, and/or the Office for Human Research Protections), or other domestic or foreign government bodies if required by law and/or necessary for oversight purposes.
   b. WIRB
   c. Outside individuals or entities that have a need to access this information to perform functions relating to the conduct of this research such as data storage companies.

   Some who may receive your protected health information may not have to satisfy the privacy rules and requirements. They, in fact, may share your information with others without your permission.

5. **For how long will protected health information about me be used or shared with others?**
   There is no scheduled date at which your protected health information that is being used or shared for this research will be destroyed, because research is an ongoing process.

6. **Statement of privacy rights:**
   a. You have the right to withdraw your permission for the Global A-T Family Data Platform to use or share your protected health information. We will not be able to withdraw all the information that already has been used or shared with others. To withdraw your permission, you must do so in writing by contacting the researcher listed on the first page of this form.
   b. If you cancel your authorization, you will not be able to continue in the study. You can cancel your authorization for the optional part(s) of the study and remain in the main study.
   c. You have the right to request access to your protected health information that is used or shared during this research and that is related to your treatment or payment for your treatment. To request this information, please contact your doctor who will request this information from the research directors.

_____ _____

<<Electronic>> Signature of Participant or Legally Authorized Representative | Date

S. Documentation of Consent

Consent and Assent Instructions:

Consent: Subjects able to provide consent must sign on the subject line below
Subjects 18 years and older must sign on the subject line below
Consent is provided by the Legally Authorized Representative for adult subjects unable to consent
For subjects under 18, consent is provided by the parent or guardian
For subjects who are under 18 when they begin participation and become 18 during the study, consent to continue must be provided by the subject at that time using the section below

Assent: Is not required for subjects 6 years and younger and for adults with diminished capacity who require a legally authorization representative.
Written assent is required for subjects ages 7 through 17 years using the Assent Form for Children.

This is what I agree to:

- Researchers may use my health information, medical information, the results of the DNA sequencing (as indicated in the checkboxes below) and any other data I share via this platform for future research studies - including studies that have not yet been designed; studies for A-T; studies involving diseases other than A-T, and/or studies that may be for commercial purposes, subject to review and approval by the A-T data access committee. This information will be stored without identification information on a cloud-computing storage system administered by Broad Institute.

- Researchers may share this information with qualified researchers in a manner that does not include my name, personal identifying number (such as social security number in the United States, or Health Insurance Number in Canada), or any other information that could be used to readily identify me, to be used by other qualified researchers to perform future research studies, including studies that have not yet been designed, studies for A-T; studies involving diseases other than A-T, and studies that may be for commercial purposes.

Check the boxes below to indicate if you agree to the following options. If you check "no" to any given option, you can still take part in the initiative.

YES	NO	
☐	☐	Study staff may contact me with follow-up research questionnaires and invitations to participate in additional studies. I may choose to ignore these questionnaires/invitations.
☐	☐	Study staff may perform (or collaborate with others to perform) DNA sequencing on the saliva sample that I will send them and store the sample until genomic sequencing is successfully completed.
☐	☐	Study staff may request my medical records from my physicians and hospitals.
☐	☐	Study staff may contact me in the event that it becomes possible to return genetic results to my physician or me.

☐ ☐ Study staff may contact my physician if a researcher reports genetic analysis results about my A-T mutations.

My signature below indicates:

- I have had enough time to read the consent form and think about agreeing to participate (or allowing my child to participate) in this study.
- I have had all of my questions answered to my satisfaction.
- I am willing to participate (or to allow my child to participate) in this research study.
- I have been told that my (or my child's) participation is voluntary and if I decide not to participate it will have no impact on my (or my child's) medical care.
- I have been told that if I decide to participate now (or allow my child to participate), I can decide to stop being in the study (or withdraw my child from the study) at any time.
- I agree to allow the collection, use, and sharing of my (or my child's) information as described above.
- By signing this form, I do not give up any of my (or my child's) legal rights.
- You should print a copy of this document for your records.

_____
Printed Name of Participant

_____
Date

If participant does not have the legal capacity to consent to their participation:
I am the parent/guardian or legally authorized representative of the participant named above and I consent to his/her participation in this research study. I also authorize the collection, use and sharing of the participant's information.

_____
Printed Name of Parent/Guardian
or Legally Authorized Representative

_____
Date

I attest that the individual providing consent had enough time to consider this information, had an opportunity to ask questions, and voluntarily agreed to participation in this study.

_____
Printed Name of Person Explaining Consent

_____
Date

WITNESS STATEMENT
As an impartial third party, I witnessed the entire consent discussion and the signature of the individual providing consent on this form.

_____
Printed Name of Witness

_____

Date