

APPENDIX A

Adult Participant Statement of Preferences on Return of Results to Family Members and Designation of a Representative

Points to Consider

- **Timing.** This form can be incorporated into the initial consent process or be administered at a later time. The research team should decide when it is most appropriate to discuss and provide this form.
- **Communication process.** This form should be part of a larger communication process between the research team and the participant. The form can be used to facilitate that conversation and document the participant's views on sharing and who should serve as the participant's Representative.
- **Views on sharing results.** This form asks simply whether the research participant wants their research results shared with family members or not. Some research studies may choose to ask more detailed questions, such as what kind of results should be shared and with which family members.
- **Key elements.** This document should explain that relatives may be interested in the participant's genetic research results, say how family members' requests for results will be handled, explain the need and importance of a Representative and allow the participant to designate a preferred Representative, and elicit the participant's preferences for sharing research results with family members.

Suggested Form

Research Participant's Name: _____

Date of Birth: _____ Phone #: _____

This study involves genetic testing [OR GENOME SEQUENCING]. Your genetic information is unique to you, but you share some genetic similarities with your blood relatives, including your children, parents, brothers, and sisters (when they are biologically related to you). You may wish to consider whether you want to share your genetic results with family members, so they can decide whether to be tested to find out if they have the same result.

Please note that there is no guarantee that your family members will get any benefit from receiving the results. There is a risk that they may feel distress from learning the result. There is also a risk that our understanding of the result might change as we learn more.

You are free to tell your family members about your results at any time. If you share your results with family members, there is a risk that they will share your results with other people.

If your family members decide to ask the research team about your genetic results, **we will ask them to talk to you** so you can decide whether they should get your result. If you can no longer decide for yourself (for example, because you are too sick to decide or are deceased), we would like to refer them to someone you trust — your Representative — to decide about access to your results.

Please name below the person you would like to be your Representative. This may be a family member or someone else you trust. You may want to give a copy of this form to your Representative and talk about your preferences for sharing research results with family members.

Please note that there are some cases in which a family member has a legal right to a relative’s genetic information, even if the Research Participant or Representative objects. We would be happy to answer any questions you have about this.

In rare cases, we may identify a genetic result that we would like to share with a family member to prevent harm to their health. This would be a genetic result that your family member is likely to have too and that poses a high risk of imminent health harm to them. If the high risk can be reduced by telling them so they can get genetic testing, we would like to share the result with them. In that case, we will first recommend that you (or your Representative, if they are making access decisions at that point) reach out to the family member. However, in these rare cases, we may reach out directly to the family member ourselves to prevent harm, even if you or your Representative objects.

A. Your Preferred Representative: I would like the person named below to decide on family access to my genetic results if I am unable to decide for myself.

Representative’s Information:

Name: _____

Relationship to you: _____

Address: _____ City: _____ State: _____ Zip: _____

Telephone: _____ Cell: _____ Email: _____

B. Your Preferences on Offering Results to Family — Guidance for your Representative: I authorize my Representative to share my genetic results with family members (initial your choice):

Yes _____ No _____

Do you have any further guidance for your Representative on your preferences?

.....
Your rights: I understand I have a right to:

- **Ask questions** about this form
- **Think** about this form and wait before signing it
- **Refuse to sign** this form
- **Receive a copy** of this signed form
- **Change my mind** and revoke or change this form at any time
- **Continue** as a research participant in the study, whether I sign this form or not.

.....
Signatures:

The research team has discussed these choices with you and answered your questions. By signing this page, you acknowledge that you have read and agreed to the terms of this form:

Signature of Research Participant: _____ **Date:** _____

Printed name of Research Participant: _____

Signature of Individual Obtaining Preferences: _____ **Date:** _____

NOTE: If you have any questions or concerns about this form, please contact [INFORMATION].

APPENDIX B

Guidance Letter for Representative

Points to Consider

- **Timing.** The research team may provide this letter once the participant has named his/her preferred Representative (Appendix A) or may provide it at a later time. The research team should decide what timing is most appropriate.
- **Recipient.** The research team may choose to provide this letter to the participant who can then give it to the Representative. Alternatively, the research team may send this letter to the Representative directly.
- **Key elements.** This document should include general background about the research study, notify the Representative that he or she has been named by the participant as their preferred person to serve as Representative, outline the process by which family requests for results will be handled, provide substantive guidance on the participant's sharing preferences, alert the Representative to the possible disclosure of results under other Federal and State laws, and provide contact information.

Suggested Form

Dear [REPRESENTATIVE]:

Date: _____

[RESEARCH PARTICIPANT'S NAME] is a Research Participant in [STUDY]. [INCLUDE brief description of research study]. The Research Participant has requested that you serve as his/her Representative to make decisions about others' access to his or her genetic results if he/she can no longer make decisions. This means that if the Research Participant loses the capacity to make these decisions or dies, you may be asked to make these decisions instead.

If the Research Participant's family member(s) seeks research results and the Research Participant can no longer make decisions about this, **the family member(s) will be referred to you** to make decisions about their access to the genetic results.

We asked whether the Research Participant wished to share results with family member(s). The Research Participant said that he/she DID/DID NOT [INDICATE WHICH ONE] wish to share. We also asked whether the Research Participant had any more preferences on sharing results with family members. The Research Participant indicated NO ADDITIONAL PREFERENCES/THESE ADDITIONAL PREFERENCES: _____ . [INDICATE WHICH ONE]

If a family member requests access to the Research Participant's results, and if the Research Participant has lost decisional capacity or died and so can no longer make decisions him- or herself, we will ask you to decide on family member access to results. You will need to consider any preferences the Research Participant expressed.

- If the Research Participant agreed to sharing results with family members, this provides strong grounds for you to permit access.
- If the Research Participant stated a preference not to share, this provides strong grounds for you to refuse access.
- If the Research Participant was silent on sharing results with family members, you should balance his/her privacy and personal interests against the interests of relatives in receiving the genetic results.

Please note that there are some cases in which a family member has a legal right to a relative's genetic information, even if the Research Participant or Representative objects. We would be happy to address any questions you have about this.

In rare cases, we may identify a genetic result that we would like to share with a family member to prevent harm to their health. This would be a genetic result that the family member is likely to have too and that poses a high risk of imminent health harm to them. If the high risk can be reduced by telling them so they can get genetic testing, we would like to share the result with them. In that case, we will first work with the Research Participant to decide on family member access. However, if the Research Participant is decisionally incapacitated or dead, we will work with you to reach out to the family member. In these rare cases, we may reach out directly to the family member to prevent harm, even over the objection of the Research Participant or you as the Representative.

Serving as the Representative is an important responsibility. If you have any questions or concerns, please contact [INFORMATION].

Signed,
[RESEARCHER]

APPENDIX C

Consent Form for Family Member to Receive Participant's Research Results

Points to Consider

- **Applicability.** The consent form for family members may be useful in several scenarios: (1) the participant asks the research team for help in returning the research results to family members; (2) the participant's Representative asks the research team for help in returning the research results to family members; or (3) in the rare circumstance in which the research team reaches out directly to family members to offer the result in order prevent imminent harm. When the Representative is sharing research results with family members without the involvement of the research team, the Representative may prefer a more informal approach that does not utilize this form.
- **Genetic counseling.** Some research teams may provide access to a genetic counselor for family members; other teams may instead refer family members to a genetic counselor. Research teams should consider clarifying in this form whether they will provide access to genetic counselors and who will pay for the genetic counseling.
- **Rare circumstances.** In the rare circumstance in which researchers discover a highly pathogenic and actionable variant that a relative is likely to carry and whose disclosure is highly likely to avert imminent harm, then this consent form can be provided to the relative, who can then decide if they would like to receive the result.
- **Key elements.** This document should include background information on the research study, alert the family member to potential risks and benefits of receiving their relative's genomic research result, allow the family member to consent or refuse to receive the research result, address access or referral to a genetic counselor, and provide contact information.

Suggested Form

Your family member [NAME] has participated in a genomics research study. We are requesting your permission to return a research result to you that was found in the course of that research study. This research result will

show that your family member has a particular genetic variant. It is possible that you share this same genetic variant, but it is also possible that you do not. To be sure, you may want to get your own genetic testing.

Learning about your family member’s genetic test result may raise concerns and be upsetting. Because some genetic variations can help to predict future health problems for you and your relatives, this information might be of interest to health care providers, life insurance companies, employers, and others. However, Federal and State laws provide some protections against discrimination based on genetic information. For example, the federal Genetic Information Nondiscrimination Act (GINA) makes it illegal for health insurance companies, group health plans, and many employers to discriminate against you based solely on information about your genetic risk. However, it does not prevent companies that sell life insurance, disability insurance, or long-term care insurance from using genetic information as a reason to deny coverage or set premiums. The law in your state may or may not provide additional protections.

If you agree to receive your family member’s genetic result, please return this form with your signature to: [ADDRESS]. If we receive your agreement, we will communicate the research result to you. At your request, we will also provide [OR REFER YOU TO] genetic counseling to help you understand the result.

A. Your decision about whether to receive your family member’s genetic result:

[please check the one you choose]

- Yes, I wish to receive the result _____
- No, I do not wish to receive the result at this time _____
- I would like to speak to a genetic counselor before I decide _____

B. Your decision about whether to request genetic counseling about the result:

[please check the one you choose]

- Yes, I would like to speak to a genetic counselor about this result _____
- No, I do not wish to speak to a genetic counselor about this result at this time _____

Name: _____

Address: _____

City: _____ State: _____ Zip: _____

Telephone: _____ Cell: _____ Email: _____

Signatures:

By signing this page, you acknowledge that you have read and agreed to the terms of this form:

Signature of Family Member: _____ **Date:** _____

Printed name: _____

Signature of Individual Obtaining Preferences: _____ **Date:** _____

NOTE: If you have any questions or concerns about this form, please contact [INFORMATION].

APPENDIX D

Letter to Family Member to Share or Confirm Research Results

Points to Consider

- **Share or confirm results.** This letter can be used by either the participant or the participant's Representative to share results for the first time or to confirm results that have already been shared by the participant, Representative, researcher, or genetic counselor.
- **Genetic counseling.** Some research teams may provide access to a genetic counselor for family members; other teams may instead refer family members to a genetic counselor. Research teams should consider clarifying in this form whether they will provide access to genetic counselors and who will pay for the genetic counseling.
- **Rare circumstances.** Researchers may use this form when reaching out to family members directly to return a genetic result. We urge that researcher-initiated return be restricted to return of a highly pathogenic and actionable variant that a relative is likely to carry and whose disclosure is highly likely to avert imminent harm. In such cases, the family member should first be offered an opportunity to consent to receiving the result or to refuse receipt of the result (Appendix C). Appendix D is relevant only when the family member has consented to receive the result. Appendix D can then be used to communicate or confirm the result.
- **Key elements.** This document should provide background information on the study, the result itself, a description of the result, and the potential implications of this result. This form should explain the possibility that the relative may share the same genetic variant as the participant, but should caution the relative not to act on this result until he or she has been personally tested and undergone genetic counseling.

Suggested Form

Dear [RELATIVE]:

Date: _____

Your relative [PROBAND'S NAME] has participated in a genetic research study. In that study, we discovered that your relative has a genetic result called [NAME OF RESULT]. [INSERT description of pathogenic result and associated risk]

It is possible that you may have inherited the same genetic result. But it is also possible that you did not inherit this variant. You may want to seek genetic counseling and consider genetic testing to find out.

To arrange a meeting with a genetic counselor, please contact [INFORMATION].

If you have any further questions or concerns, please contact [INFORMATION].

Signed,
[RESEARCHER]

APPENDIX E

Guidance for Parents/Guardian Considering Family Access to Child's Research Results

Points to Consider

- **Timing.** The research team can provide this letter to the parents/guardian at the child's initial enrollment in the study or later. The research team should decide when it is most appropriate to discuss and provide this form.
- **Communication process.** This form should be part of a larger communication process among the research team, the parents/guardian giving permission for the child's participation in research, and the child (or adolescent) participant if capable of assent. The form can be used to facilitate consideration of and planning concerning questions of sharing the child participant's results with relatives.
- **Representative.** This form assumes that in most cases the parents/guardian will be serving as the child's Representative.
- **Key elements.** This document should include background on the research study and say how family requests for results will be handled, including how the child's sharing preferences will be considered when determining whether results should be shared with family members.

Suggested Form

Dear [PARENTS/GUARDIAN'S NAME]:

Date: _____

[CHILD'S NAME] is a Research Participant in [STUDY]. [INCLUDE brief description of research study]. As the child's parents/guardian, you will serve as the child's Representative. As the Representative for the child, your permission will generally be needed to share the child's research results with family members.

Your child may or may not have signed an assent form stating his/her wishes about sharing results. If your child has signed this form, we will give that to you. You may also want to talk with the child about the question of sharing research results with family members, the child's wishes, and his/her reasons for wanting to share or not.

Please note that there is no guarantee that family members will get any benefit from receiving the results. There is a risk that they may feel distress from learning the result. There is also a risk that our understanding of the result might change as we learn more.

If a family member seeks the child's genetic results and the child is a minor (under 18 years old) and is alive, **we will refer the family member to you and to the child (if he/she is capable of assenting)** to decide on sharing.

If you and the child agree to share, the child's results will be provided to you and you can then share that information with the family member.

If you and the child agree not to share, the results will not be shared.

If you and the child disagree on sharing, you should balance the child's privacy and personal interests against relatives' interest in access to the child's genetic results. You should strongly consider any well-informed preferences expressed by the child. Ultimately, we will defer to your decision on access to the child's genetic results. If the child is incapable of providing guidance, you should balance the child's interests against the relative's interests.

When the child turns 18, he/she may decide independently whether family members can have access to his or her genetic results.

Note that researchers currently debate whether they should look for or disclose genetic results that will not affect the health of the child but may predict the child's risk of getting a disease later as an adult. For this study, the researchers will decide whether to return results that may predict adult-onset diseases.

Please note that there are some cases in which a family member has a legal right to the child's genetic information, even if the child or parents/guardian objects. We would be happy to answer any questions you have about this.

In rare cases, we may identify a genetic result that we would like to share with a family member to prevent harm to their health. This would be a genetic result that your family member is likely to have too and that poses a high risk of imminent health harm to them. If the high risk can be reduced by telling them so they can get genetic testing, we would like to share the result with them. In that case, we will first work with you and the child (if he/she is capable of assenting) to reach out to the family member. However, in these rare cases, we may reach out directly to the family member to prevent harm, even over the objection of you and the child.

Signed,
[RESEARCHER]

APPENDIX F

Child/Adolescent Participant Statement of Preferences on Return of Results to Family Members

Points to Consider

- **Timing.** This form can be incorporated into the initial assent process, or be administered at a later time. The research team should determine when this is most appropriate.
- **Communication process.** This form should be part of a larger communication process among the research team, the child participant, and the parents/guardian. The form can be used to facilitate that conversation and document the participant's views on sharing research results with family members.
- **Key elements.** This form should explain that family members may be interested in the child's genetic research results, say how family requests for results will be handled, and elicit the child's preferences for sharing research results with family members.

Suggested Form

Research Participant's Name: _____ Date of Birth: _____

Our bodies contain genes — the body's instructions for how to grow and develop. Some genes can cause health problems. Our genes come from our biological parents. We also share some genes with other biological relatives, such as brothers and sisters. Some of your family members may want information about your genes, to learn about genes they might have too. If they learn about your genes, they will usually need to have their own genetic testing to know for sure whether they have the same gene.

Here are some more things to think about: Even if a family member gets your genetic result, it might not help them. There is also a risk that they may get worried or distressed if they learn the result. And our understanding of what the result means might change, once scientists learn more about it.

If a family member wants information about your genes and you are younger than 18 years old, **you and your parents/guardian will decide** about sharing that information. You and your parents/guardian may say yes or no to sharing with a family member. If you and your parents/guardian disagree, you will need to talk together to see if you can agree. If you can't agree but your parents/guardian still think the information should be shared, they can offer the information to the family member because you are under 18 years old.

Once you are 18 years old, you may decide independently whether your family member can have the information. You will be able to say yes or no.

We want you to know that in some situations a family member may have a legal right your genetic information, even if you or your parents/guardian objects. We would be happy to answer any questions you have about this.

In rare cases, we may identify a genetic result that we would like to share with a family member to prevent harm to their health. This would be a genetic result that your family member is likely to have too and that poses a high risk of imminent health harm to them. If the high risk can be reduced by telling them so they can get genetic testing, we would like to share the result with them. In that case, we will first work with you and your parents/guardian to reach out to the family member. However, in these rare cases, we may reach out directly to the family member ourselves to prevent harm, even if you or your parents/guardian objects.

My Wishes for Sharing My Results:

I would like my genetic results shared with my family members (initial your choice):

Yes _____ No _____

Do you have more advice for your parents/guardian on what you want? _____

.....
Your rights: I understand I have a right to:

- **Ask questions** about this form
- **Think** about the form and wait before signing it
- **Talk** to my parents/guardian before signing this form
- **Say “no”** and refuse to sign this form
- **Get a copy** of this signed form
- **Change** this form at any time
- **Keep going** as a participant in the study, whether I sign this form or not.

.....
Signatures:

The research team has discussed these choices with you and answered your questions. By signing this page, you are saying that you have read and agreed to the terms of this form:

Signature of Research Participant: _____ **Date:** _____

Printed name of Research Participant: _____

Signature of Individual Obtaining Preferences: _____ **Date:** _____

NOTE: If you have any questions or concerns about this form, please contact [INFORMATION].