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# Consent Addendum and Questionnaire for Return of Genetic Results

**Study Title:** Insert Full Title of the Protocol

Version Date: Month, Day, 20XX

Consent Name: Questionnaire for Return of Genetic Results

**Principal Investigator:** Investigator Name Telephone: (xxx) xxx-xxxx

You, or your child have agreed to take part in XXXXX. As part of that study we will perform genetic testing. This form allows you to choose the types of results that you would like to receive. This form should only be completed after reviewing and signing the XXXXX main study consent form.

If there is anything in this form you do not understand, please ask questions. Please take your time. You do not have to take part in this study if you do not want to. If you take part, you can leave the study at any time.

In the sections that follow, the word “we” means the study doctor and other research staff. If you are a parent or legal guardian who is giving permission for a child, please note that the word “you” refers to your child.

## Information about Return of Results

Only results verified in a CLIA-certified lab will be returned to you. A CLIA-certified lab means that the test results may be used for clinical purposes.

Also, we will not release results that are inconclusive or of uncertain significance. Only results that are of known significance will be released.

###  (Use this section subjects who have a disease/condition) What results can you expect to receive from this study?

This research will look for alterations or changes in your genes that may be associated with your primary diagnosis.

* In doing this, we may also identify incidental findings that are unrelated to your primary diagnosis.
* Incidental findings could tell us about other diseases that you may have or that you might develop, as well as the potential risks of having a child with a specific genetic disease.

### (Use this section for those subjects without a disease/condition – healthy controls) What results can you expect to receive from this study?

Your sample may be studied to identify alterations or changes in your genes that put you at increased risk of having future medical problems. For example, being a carrier for certain conditions or having different reactions to certain medications or anesthesia.

### (Use this section when appropriate)

### What results will you not receive from this study?

You will not receive results about paternity. We will not inform you if your father is not your biological father.

### Will all of my genetic variations and changes be identified?

We cannot guarantee which specific variants will be identified.

* Not all genetic alterations or risk for disease can be identified with current sequencing technology. Science is advancing rapidly. More alterations are identified each year.
* There may be genetic alterations that will not be identified or reported.
* It is possible that this study will be unable to identify the cause of your condition or other health problems. (only use if the subjects are enrolled because they have a disease or condition)
* Failure to identify a result will not exclude the possibility that you have an unidentified alteration in a gene/s or a risk of developing disease in the future.

### How and when will study results be verified and returned to you?

When the analysis is nearing completion or is completed, a member of the study team or one of your CHOP healthcare providers (if you allow us to contact them) will contact you to set up a time to discuss the results.

Your results will be made available to you as part of a genetic counseling session at CHOP.

Sometimes the meaning of the results will be uncertain. It is important to know that our understanding of genetics is changing quickly, and in many cases we will not know for sure what the results mean for your future health.

Sometimes, even if you learn of a clear diagnosis, there will be no specific treatment.

## Result Options

Receiving results is completely voluntary; you may choose the types of information you wish to receive. You will only receive results that are immediately medically actionable.

**Please indicate below the types of results you would like to receive by circling your choice and initialing in the appropriate box for each result type.** You will be able to change your mind about your choices prior to receiving results. Please note that the choices made on this form only apply to results from research testing and reanalysis, and do not affect results you may receive from on-going clinical testing.

\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ **Date Completed**

## Overall Result Options

We will not contact you about research results from your samples unless given permission to do so. Please note that we will only contact you about results that have important health implications, have available interventions, and have been clinically verified by a valid clinical test.

\_\_\_\_\_\_\_\_\_\_\_ (initials) I would like to be contacted about genetic test results.

\_\_\_\_\_\_\_\_\_\_\_ (initials) I do not want to be contacted about genetic test results.

**What does it mean if I am not contacted?**

For many participants, only certain genes will be analyzed, so we will not find all gene variants that cause disease. You should not assume that if you are not contacted, that you do not have any gene variants that might be related to a disease.

Not all genes will be studied in every participant; you may have a change in a gene that was not studied. Knowledge about the importance of a gene can change over time. You may have a gene variant that was not considered important at the time it was discovered. This may change if more information is learned about that gene. Results will not be re-analyzed in this current project.

# Choices for Types of Results that can be returned

Indicate your choices in the sections below.

## Result Options Related to Your Diagnosis (Only if the study targets individuals with a specific disease or condition; otherwise delete)

|  |
| --- |
| **Results Of Known Significance Related To Primary Diagnosis** |
| You will receive these results automatically. | You will receive results that pertain to your main symptoms or diagnosis.These results would include genetic alterations that have a clear effect directly related to your main medical problem or diagnosis (be specific). |

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| **Results Of Uncertain Significance Related To Primary Diagnosis** |
| You will not receive these results. | You will not receive results that are of uncertain significance related to your primary diagnosis*.* Genetic testing often identifies results that we do not know how to interpret. Some alterations do not have a clear meaning, either because they have not been seen before or because they are not well understood. Example: We may identify a new genetic alteration that have never been seen before in a gene known to be associated with your primary diagnosis. Because the alteration has never been reported before we cannot be totally certain that it is associated with disease, so it would have uncertain significance.  |
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## Result Related to Childhood Conditions or Diseases

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| **Results That Are Immediately Actionable** |
| YesNo(Initials) | You will receive results that are immediately actionable. Actionable means that:* The results have a known scientific validity and significance;
* Knowing the results would have an immediate impact on your medical care.

These results would include genetic alterations that cause a condition with an immediate, significant and long-term impact on your health and/or healthcare based purely on the genetic test results and unrelated to onset of symptoms. Example: There are a limited number of rare diseases with immediate medical actionability. Some examples include conditions that cause a predisposition to certain cancers, such as *retinoblastoma* (cancer of the eye), or those with a strong predisposition to heart disease, such as *Long QT syndrome* and *Marfan* *syndrome*. |
|  |
| **Results Related to Diseases with Onset in Childhood and That Could Be Medically Actionable**  |
| YesNo(Initials) | **Results Related To Medically Actionable Childhood Onset Disease** *I want to receive results that are proven to be medically actionable and related to a disease that can have an onset in childhood.*These diseases may have variable symptoms or age of onset; an individual may not develop all or any symptoms even though they carry a genetic alteration that can cause the disease. It is also possible that a person may carry an alteration but not yet have symptoms or have symptoms that have not yet been recognized. Results related to diseases that have an immediate impact on your health or healthcare will be automatically released.This category includes any childhood onset disease that is medically actionable and known to cause symptoms prior to the age of 18. Medically actionable results include genetic alterations that: 1) clearly affect the functioning of a gene known to be associated with disease; AND2) could cause a serious risk to your health; AND3) there are known options for improving your health or remaining healthier through changes in how you and your healthcare providers treat your condition or manage your health care. Example: There are a large number of childhood onset diseases with various health issues. Some examples of childhood onset genetic diseases include certain types of *ichthyosis* (dry and itchy skin) and *albinism* (decreased vision and decreased color of the hair and skin). |
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## Results Related To Your Health In Adulthood

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| **Results Related to Medically Actionable Adult Onset Disease** |
| YesNo(Initials) | *I want to receive results that are of proven medical importance. This means that the results are medically actionable and related to a disease with onset in adulthood.* Knowing about these conditions can allow a person to undergo increased screening and preventive measures and improve your long-term health. Medically actionable results include genetic alterations that: 1. clearly affect the functioning of a gene known to be associated with disease; AND
2. could cause a serious risk to your health; AND
3. have known options for improving your health or remaining healthier through changes in how you and your healthcare providers treat your condition or manage your health care.

Example: Examples of results that are medically actionable include finding a change in a gene known to *cause polycystic kidney disease (*causes hypertension, aneurysms and kidney problems) or making you more likely to get *cancer*. Knowing about these diagnoses would allow someone to improve symptoms or avoid serious health problems through medication, special monitoring or other preventive measures, making this information medically actionable.  |
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| **Results of Carrier Status for Recessive Disorders** |
| YesNo(Initials) | *I want to receive results of carrier status for recessive diseases.* Knowing carrier status can allow a family to make informed decisions about family planning. Only results that are pathogenic or likely pathogenic will be returned in this category (variants of uncertain significance (VUSs) will not be returned. Every person carries recessive alterations in genes known to be associated with human disease. * Recessive diseases are most often caused by a child inheriting an alteration in the same gene from both parents.
* Carriers of an alteration in only one copy of a recessive gene do not generally have symptoms.
* If both parents carry an alteration in the gene, then each of their children has a 25% (1 in 4) chance of inheriting the disease.
* Females can also be carriers of recessive disorders caused by alterations in genes on the X chromosome.

Example: There are many different recessive diseases, including *cystic fibrosis* (lung infections and intestinal problems), *Tay Sachs disease* (loss of muscle and brain function)*,* and *beta-thalassemia* (anemia and enlarged spleen and liver). |
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## Results Related Drug Processing and Complex Traits

Our genes can influence the way the body responds to drugs. The study of these genes is called pharmacogenetics. Sometimes several genes rather than one gene influence the development of a disease or condition. These are considered complex traits.

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| **Results of Risks for Pharmacogenetic and Complex Traits** |
| YesNo(Initials) | *I want to receive results of risks for pharmacogenetic and complex traits.* Every person carries variants in their DNA known to be associated with complex human disease and the way our bodies metabolize medications. * Complex human diseases are usually caused by changes in one or more genes in combination with environmental exposures – drugs, foods, tobacco, activity levels etc.).
* Carriers of a variant for a complex disease may have an increased risk of getting that condition but may also never get the condition.
* Pharmacogenetic traits refer to changes in DNA that may affect the way the body uses or metabolizes certain medications. These differences can make some people more sensitive or resistant to specific medications.

Example: There are many different complex diseases, including *diabetes* (high blood sugar), *hypertension* (high blood pressure)*,* and *coronary artery disease* (compromise to the blood vessels that send blood to the heart). Pharmacogenetic traits may include sensitivity or resistance to certain medications like coumadin (a blood thinner) or certain anesthetics. |
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## What about privacy and confidentiality?

The privacy and confidentiality protections outlined in the main consent form apply to the return of results.

By law, CHOP is required to protect your private information. The investigator and staff involved in the study will keep your private information collected for the study strictly confidential. Please refer to the main consent form that explains more specifically how your personal information will be protected.

## How do I submit my completed form?

Thank you for selecting your result choices. You can change your mind and complete this form again at any time. Once completed, this form should be submitted directly to your CHOP clinician/genetic counselor or mailed to the Project Coordinator:

Dr. XXXXXX
The Children’s Hospital of Philadelphia
Division/Department
34th Street and Civic Center Blvd.
Philadelphia, PA 19104

## Consent to Contact Referring Clinician

Please initial below if you are comfortable with the study team informing members of your clinical care team of your participation in this study. If your clinician is a member of the study team, they may be notified of your participation in the study without this consent.

\_\_\_\_\_\_\_\_\_\_\_ (initials) I give permission for the study team to contact my clinician regarding this study.

\_\_\_\_\_\_\_\_\_\_\_ (initials) I do not want my clinician to be contacted regarding this study.

## Consent to have Genetic Results Returned

The results options have been explained to you by:

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| --- | --- | --- |
|  |  |  |
| Person Who Explained Results Options |  | Signature of Person Who Explained Results Options |
|  |  |  |
|  |  | Date |

By signing this form, you are indicating that you have had your questions answered and that you agree to the results options selected above.

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| Name of Subject whose Results Choices are listed above |  |  |
|  |  |  |
| Signature of Subject (18 years or older) |  | Date |
|  |  |  |
| Name of Authorized Representative (if different than subject) |  | Relation to subject:[ ]  Parent [ ]  Legal Guardian |
|  |  |  |
| Signature of Authorized Representative |  | Date |

### For children (between the ages of 13 and 18 years) who assented to study participation:

The results options have been explained to me and I agree to the options selected above.

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| Signature of Subject (age 13 to 18 years) |  | Date |

If the child was unable to assent, please specify the reason why below.
(Example: cognitive impairment, sedation, etc.):