

SPECIMEN INFORMATION

Specimen:

- Blood** (5-7mL K₂EDTA/K₃EDTA)
- DNA - Source:** _____
- Other** _____

Date Collected: (mm/dd/yyyy) _____ / _____ / _____

Space for Lab Use Only

PATIENT INFORMATION

First name: _____ **MI:** _____
Last name: _____
Date of Birth: (mm/dd/yyyy) _____ / _____ / _____
Gender: Male Female Unknown/Unspecified
Address: _____

City: _____ **State:** _____ **Zip Code:** _____
Phone: _____
Email: _____

Institution: _____
Medical Record Number: _____
Is the patient adopted? Yes No
Is the patient deceased? Yes No *if yes, date:* _____
Race and Ethnicity: *Please check ALL that apply*
 White Ashkenazi Jewish Hispanic Asian
 Black/African American American Indian/Native Alaskan
 Native Hawaiian or other Pacific Islander Other _____

REFERRING PROVIDER INFORMATION

Referring Provider

Name (First, Last): _____
Phone: _____ **Fax:** _____
Email: _____
Institution: _____
Address: _____

City: _____ **State:** _____
Zip Code: _____ **Country:** _____

Genetic Counselor / Additional Contacts

Name (First, Last): _____
Phone: _____ **Fax:** _____
Email: _____
Institution: Same as Referring Provider Provided below

Place facility sticker here

PAYMENT INFORMATION

Please note: Payment information must be completed for testing to begin.

Patient Pay (please complete section in its entirety)**
 Check (please attach to forms)*
Please make checks payable to Partners HealthCare Personalized Medicine
 Credit card (please fill out credit card information in its entirety)
Card type: Mastercard Visa AMEX
Name (as it appears on card): _____
Credit card number: _____
Expiration Date: _____ **3 Digit Security Code:** _____

***For patient pay, please provide billing address and contact information.
 If same as above, please note section as such.***

Patient Pay Billing Address: _____
City: _____ **State:** _____ **Zip Code:** _____ **Country:** _____
Home: _____ **Cell/Work:** _____ **Email:** _____

Referring Institution or Research Study
 (please complete section in its entirety)
Bill to Name/Department: _____
Address: _____

City: _____ **State:** _____
Zip Code: _____ **Country:** _____
Phone: _____
Contact Person: _____

GENOME SEQUENCING REQUISITION FORM

Patient Name: _____ Date of Birth: ____/____/____ (MM/DD/YYYY)

TEST TO BE PERFORMED

Check box(es) to order test(s). Please complete a separate form for each individual to be tested.

Genome Sequencing and Interpretation

Report Incidental Findings recommended by ACMG?
 Yes No, I wish to opt out of Incidental Findings

Trio - Genome Sequencing and Interpretation

Trio Proband

Report Incidental Findings recommended by ACMG?
 Yes No, I wish to opt out of Incidental Findings

Trio Auxillary Member

Relationship to Proband _____
 Proband Name _____
 Report Incidental Findings recommended by ACMG?
 Yes No, I wish to opt out of Incidental Findings

*Expanded incidental findings analysis is available.
 Contact the laboratory for more details.*

Family Member Sample for Support of Proband Testing

(Variant confirmation only - Genome Sequencing will not be performed)
 Relationship to Patient _____
 Proband Name _____

Familial Variant Testing

(Sequencing - first 3 variants, \$500; each additional variant, \$50)

Familial Variant(s) OR Research Confirmation

Gene _____
 Variant _____
 Proband Name _____
 Relationship to Patient _____
 LMM Accession #: PM- _____

CLINICAL INFORMATION

**Detailed patient clinical history is required. Additional clinic notes may be attached to requisition form.*

Purpose of Study and Pertinent Clinical Information (provide any suspected clinical diagnoses or state if unaffected): _____

Previous Genetic Testing: No Yes - Provide results

Suspected Mode of Inheritance: Dominant Recessive X-Linked De novo Mitochondrial

Medical History: For any area marked "Abnormal" please provide additional information

ICD-10 Code (s): _____

	Normal	Abnormal	Unknown	Description
Abdomen	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Cardiovascular	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Chest/Thorax	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Endocrine	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Genital System	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Growth and Build	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Head and Neck	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Hematology	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Immunology	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Metabolic	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Muscle, Soft Tissue	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Neoplasia	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Neurologic	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Respiratory	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Skeletal	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Skin, Nails, Hair	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Urinary System	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Perinatal/Prenatal History	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Abnormal Laboratory Results	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
Other	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	

PLEASE PROCEED TO PAGE 3 TO COMPLETE THE FAMILY HISTORY

GENOME SEQUENCING REQUISITION FORM

Patient Name: _____ Date of Birth: ____/____/____ (MM/DD/YYYY)

CLINICAL INFORMATION

Please list any genes for which you are requesting specific analysis or note any special requests here. (Please note: The lab will also generate a list of genes and variants based on clinical features and purpose of study unless directed otherwise):

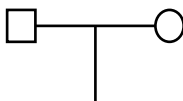
FAMILY HISTORY

Family History: Please list family members available for testing, their relationship to proband and affected status, or sketch below.

RELATIVE NAME / RELATIONSHIP	SAMPLE
Mother: Clinically Affected: <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<input type="checkbox"/> Sent <input type="checkbox"/> Available to Send <input type="checkbox"/> Unavailable
Father: Clinically Affected: <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<input type="checkbox"/> Sent <input type="checkbox"/> Available to Send <input type="checkbox"/> Unavailable
Sibling: Clinically Affected: <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<input type="checkbox"/> Sent <input type="checkbox"/> Available to Send <input type="checkbox"/> Unavailable
Sibling: Clinically Affected: <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<input type="checkbox"/> Sent <input type="checkbox"/> Available to Send <input type="checkbox"/> Unavailable
_____: Clinically Affected: <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<input type="checkbox"/> Sent <input type="checkbox"/> Available to Send <input type="checkbox"/> Unavailable
_____: Clinically Affected: <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> Unknown	<input type="checkbox"/> Sent <input type="checkbox"/> Available to Send <input type="checkbox"/> Unavailable

ADDITIONAL FAMILY HISTORY

Notes: _____



Race/Ethnicity: _____
 Paternal Ancestry: _____
 Maternal Ancestry: _____
 Consanguinity: Yes No

(Sketch above or attach pedigree, if appropriate)
 ○ = Female □ = Male ◇ = Gender Unspecified
 ● ■ ◆ = Affected Individual ⊙ = Carrier



Laboratory for Molecular Medicine

65 Landsdowne Street • Cambridge, MA 02139

Phone: (617) 768-8500 • Fax: (617) 768-8513 • Email: Imm@partners.org

<http://www.partners.org/personalizedmedicine/Imm>

**CONSENT FOR CLINICAL TESTING USING EXOME
OR GENOME (GENOMIC) SEQUENCING**

Patient Name: _____ Date of Birth: ____/____/____ (MM/DD/YYYY)

SPECIMEN & SHIPPING REQUIREMENTS

The preferred blood specimen is a 7mL blood sample (3-5mL for infants) collected in a lavender top (K2EDTA or K3EDTA) blood tube. All samples must be labeled with two unique patient identifiers, preferably the patient's full name and date of birth. Please contact the laboratory for more details and blood tube kits.

These samples must be accompanied by a requisition form, and the ordering provider and patient must sign the declaration below.

Specimens (with form) should be shipped overnight at room temperature to:

Laboratory for Molecular Medicine
65 Landsdowne Street
Cambridge, MA 02139

For more detailed information about shipping requirements and procedures, please contact the lab at 617-768-8500.

CONSENT FOR CLINICAL TESTING USING EXOME OR GENOME (GENOMIC) SEQUENCING

Please read this form carefully. You are being offered a blood test that looks at your genetic material. We hope this blood test will help us understand your medical condition better. This consent form will try to explain the risks and benefits of doing the test, the possible costs, and where the results are stored.

The consent form will also describe what is known and not known about genomic sequencing. Genomic testing is a new field and is changing quickly. A member of the health care team will help you understand this consent form, genomic testing, and what you can learn from the testing.

You should keep your health care team updated with your current contact information. This will make it possible for us to contact you if we learn important information from this testing now or in the future.

1. What is the purpose of the genomic sequencing test?

The purpose of this test is to find genetic changes that might be associated with your/your child's health and/or the following condition(s): _____

2. What will be required of me/my child to get the test?

You/your child will be asked to provide a sample for testing such as a blood sample or other tissue. You/your child will be asked to provide medical and family history information to help interpret the results of testing. After testing is completed, further medical or family history information, or further medical tests may be suggested to help interpret the results of testing.

3. What does the genomic sequencing test look for?

This test looks for genetic changes that are associated with specific diseases. This test may find changes in one or more genes that cause or contribute to your/your child's condition. The test may also find changes that make it more likely that you/your child will develop certain diseases.

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It is possible that this test will not find any specific changes that explain your/your child's condition. This result would not mean that you/your child do not have or will not develop a genetic disease.

4. What are the limits of the genomic sequencing test?

This test will find a large number of genetic changes. However, some genetic changes are not currently detectable with the testing methods used. For those detected, we will not know what many of these changes mean. It may be many years before we understand what all of the changes mean and are able to find a change that causes or contributes to your/your child's condition. Our testing process includes highly skilled technicians and advanced technology. As in any laboratory, there is a small possibility that the test will fail.

5. How will I learn the results of the genomic sequencing test?

You will learn the results of this test from: _____. This healthcare provider will receive a laboratory report that discusses any results that may be relevant to the reason you/your child had testing. Your/your child's healthcare provider will share this information with you, and a genetic counselor may assist in explaining the results.

6. How long will it take to get my results?

The Laboratory for Molecular Medicine (LMM), who will be performing this test, will make every effort to get the results to your healthcare provider as quickly as possible after receiving your sample. Please see our Genomic Sequencing FAQs (<http://personalizedmedicine.partners.org/Laboratory-For-Molecular-Medicine/FAQ/Exome-Genome-Sequencing.aspx>) for more detailed information. Listed turn-around times represent the typical turn-around time for a test, but are not guaranteed.

7. Will I get all of my results at once?

Because the understanding of genetic changes will improve over time, it is likely that more will be understood about your results after you get your initial report. If new information is discovered that is thought to be important to your health and/or the health of your relatives, your/your child's healthcare provider may be re-contacted.

In some cases, the laboratory will only look for new information in your/your child's genetic results if you or your healthcare provider requests it. If you would like an updated report, please contact the lab. There may be a charge for a new interpretation of your results.

After the initial report is generated, you or your ordering healthcare provider may request an electronic copy of your full genomic sequence data files from the LMM by calling (617) 768-8500. There may be a charge for the laboratory to provide your data to you. The laboratory may not be able to store your data indefinitely but will store your results for at least 18 months.

8. How could the genomic sequencing test affect my family members?

This test could reveal information about the health of your relatives, such as their chances of developing certain diseases. Such information could be unexpected, or it could explain a medical condition in your family.

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Patient Name: _____ Date of Birth: ____/____/____ (MM/DD/YYYY)

If the test finds a genetic change that may be important to your family's health, your/your child's healthcare provider will ask you to tell your family members about it.

This test will find many changes that we cannot interpret. In this case, testing your parents or other family members to see if they have the same change may help understand certain results. Your/your child's healthcare provider may ask you if your family members would be willing to have genetic testing. Certain followup testing may be performed free of charge.

9. What other information can the genomic sequencing test tell me?

The test may find genetic changes that tell us that you/your child are at risk for diseases other than your/your child's condition, such as cancer risk. These changes are often called "incidental" or secondary findings. If incidental findings are thought to be medically important for you/your child, they may be reported to your/your child's healthcare provider. Your/your child's healthcare provider will talk to you about how these findings may change your/your child's risk for disease and/or medical care.

For more information on incidental findings, please go to our Genomic Sequencing Frequently Asked Questions section of our website: <http://personalizedmedicine.partners.org/Laboratory-For-Molecular-Medicine/FAQ/Exome-Genome-Sequencing.aspx>

10. Will my genetic information become part of my medical record?

Your/your child's genomic sequencing report will list medically important genetic changes that were found by the genomic sequencing test. This report containing your/your child's test results as well as any updates to those results will become part of your/your child's permanent electronic medical record and be made available to any healthcare provider treating you now or in the future. Your protected health information will be used in accordance with the terms of the Partners Healthcare Privacy notice. As we understand more about genetic changes, your/your child's report and medical record may be updated.

The laboratory will not initially deposit your complete genetic sequence into your/your child's medical record. However, it is possible that this policy will change in the future in which case your/your child's complete genetic sequence may be incorporated into your/your child's permanent medical records.

11. How will my blood sample and genetic information be used?

After running your test, the laboratory may use your/your child's remaining sample to do quality control checks and/or to develop new and better laboratory tests. Your/your child's name and other identifying information will be removed from the sample before it is used in these ways. In addition, your de-identified sample may be used in accordance with the research policies stated in the following section. There will be no additional charge to you for using your/your child's sample in these ways.

12. What are the risks of the genomic sequencing test?

You may learn medical information about yourself/your child that you did not expect. Learning that you/



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your child are at risk for a disease other than your/your child's condition, and which may or may not be preventable or treatable, could lead to emotional or psychological distress.

You may discover things about yourself/your child that trouble you and that you may not have the ability to control or change. For example, unexpected parental relationships or other information about your ancestry could be uncovered.

Your relatives may be upset to learn that they may be at risk for a disease.

The test will give us a lot of information, but we won't know what all of it means right away. It is possible that this test will not find the cause of your/your child's condition, or the test may not tell us your/your child's chance of developing specific diseases. This could be frustrating or upsetting.

Although there are laws to prevent employment and health insurance discrimination based upon genetic findings, there are currently no laws to prevent the use of genetic information on the ability to obtain life, disability or long-term care insurance.

13. Will anyone else have access to my genomic sequence, shared medical history or interpreted results?

The ordering physician can obtain access to your genomic sequence data files for the purpose of your clinical care.

Test results and submitted clinical information may be shared with other clinical laboratories for the purpose of improving our understanding of the relationship between genetic changes and clinical symptoms. Sharing data in this manner may enable us to provide better interpretations of your genetic findings as well as assist other patients with similar results. We will protect your privacy/confidentiality by replacing your name and other direct identifiers, such as date of birth or medical record number, with a code. The key to the code numbers will be stored securely in the testing laboratory. We will share only de-identified information with outside clinical labs.

14. Who besides my physician can I talk to if I have more questions about the genomic sequencing test?

You may request the name and phone number of a genetic counselor who can answer your questions about the genomic sequencing test and help you understand your/your child's test results.

15. Will I have to pay for the genomic sequencing test?

Your health insurance may or may not cover the cost of this test. You will need to pay any portion of this test that is not covered by your health insurance. You may also have medical visits to follow up on the results of this test. Insurance coverage for such tests will be subject to current medical practice and your insurance policy coverage. If you seek insurance coverage for this test, you may be required to release your/your child's results to your health insurance company for payment purposes.



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Patient Name: _____ Date of Birth: ____/____/____ (MM/DD/YYYY)

RESEARCH POLICIES & OPPORTUNITIES

Blood or other samples sent to the Laboratory for Molecular Medicine (LMM) may be used by Partners Healthcare System (PHS), by medical organizations connected to PHS, or by educational or business organizations approved by PHS, for research, education and other activities that support PHS's mission, without your/the patient's specific consent. Other types of research performed in association with the LMM require that we obtain consent from the patient (see below).

PATIENTS - Please check off and initial below whether we can contact you to let you know about research studies requiring consent in which you/your child may be able to participate. These research studies may include:

- A request for additional clinical records about your condition
• Studies to find new causes for your condition
• Studies to evaluate newly developed treatments for your condition

Please check one option:
[] Yes, you can contact me _____ (patient initials)
If yes, please provide your contact information on the first page
[] No, please do not contact me _____ (patient initials)

RESEARCH POLICIES & OPPORTUNITIES

I have carefully reviewed the above. I have had my questions answered to my satisfaction. I consent to provide a sample for testing by genomic sequencing.

_____ Date _____ Time _____ AM/PM
Patient/Surrogate Decision Maker Signature

_____ Patient Name (Print) _____ Surrogate Decision Maker Name if applicable

ORDERING PROVIDER SIGNATURE

I, _____ (print name), as ordering provider, certify that I understand and accept that the patient being tested and/or their legal guardian have been informed of the risks, benefits, expectations and limitations of the testing ordered, as well as the policies of the LMM listed above. I have obtained informed consent, as required by my own state and/or federal laws. In addition, I assume responsibility for returning the results of genetic testing to my patient and/or their legal guardian and for ensuring that my patient receives appropriate genetic counseling to understand the implications of their test results.

____ Other
NP
PA

Signature _____ MD CID _____ Date _____ Time _____ AM/PM