Mass General Brigham

Personalized Medicine
Laboratory for Molecular Medicine
65 Landsdowne Street • Cambridge, MA 02139
Phone: (617) 768-8500 • Fax: (617) 768-8513
The LMM is a satellite facility of Massachusetts General Hospital. • CLIA # 22D1005307

SPECIMEN INFORMATION

Specimen: ❑ Blood **Date Collected:** (mm/dd/yyyy) _____ / _____ / _____
❑ Saliva *(NOT accepted for panel testing, only for Familial Variant Testing.)*
❑ Cord Blood *(Maternal cell contamination studies must be completed prior to sending and report must be attached to this form.)*
❑ DNA derived from (*Choose One*):
❑Whole Blood ❑ Cord Blood* ❑ CVS* ❑ Amnio* ❑ Other_____
If proving DNA, name and CLIA # of lab performing blood draw: _____

PATIENT INFORMATION

First name: _____ MI: _____
Last name: _____
Date of Birth: (mm/dd/yyyy) _____ / _____ / _____
Gender: ❑ Male ❑ Female ❑ Unknown/Unspecified
Is patient pregnant? ❑ No ❑ Yes EDD:_____
Address: _____
City: _____ State: _____ Zip Code: _____
Phone: _____
Email: _____

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

❑ **Referring Institution** (please complete section in its entirety)
For new referring facilities, please complete and submit the New Institution Add Form
Bill to Name/Department: _____
Address: _____
_____
City: _____ State: _____
Zip Code: _____ Country: _____
Phone: _____
Contact Person: _____

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

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

SPECIMEN & SHIPPING REQUIREMENTS

The preferred blood specimen is a 7 ml blood sample (3-5ml for infants) collected in a lavender top (K_2EDTA or K_3EDTA) blood tube. Smaller blood samples or other tissue specimens may also be acceptable for certain tests. All samples must have two patient identifiers, preferably the patient's name and date of birth. Please contact the laboratory for more details.

Each sample must be accompanied by a requisition form (available at Partners.org/PersonalizedMedicine/Laboratory-For-Molecular-Medicine/Ordering). The ordering provider must sign the declaration below.

The blood sample (with forms) 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, see our website Partners.org/PersonalizedMedicine/Laboratory-For-Molecular-Medicine/Ordering/Sample-Requirements-Payment-Shipping.

LABORATORY FOR MOLECULAR MEDICINE POLICIES

By requesting testing from the Laboratory for Molecular Medicine (LMM), the ordering provider indicates that he/she understands AND accepts the policies of the LMM, as noted below, and has communicated these policies to the patient.

1. Our testing process includes highly skilled technicians and advanced technology. As in any laboratory, there is a small possibility that the test will not work properly, or an error may occur.
2. Listed turn around times (TATs) represent the typical TAT for a test, but are not guaranteed.
3. If the requisition form is incomplete, and the healthcare provider cannot provide the required information, lab staff may need to contact patients directly to obtain or verify the information needed to complete the form.
4. Test results, as well as any updates to those results, may become part of a patient's permanent medical record (electronically or otherwise) or be made available (electronically or otherwise) to the ordering healthcare institution and its healthcare team.
5. Results will only be released to the ordering provider and other providers listed on the requisition form. The ordering provider assumes the responsibility to disclose the test results and direct care as appropriate.
6. The ordering provider can obtain access to your genomic sequence files for the purpose of your clinical care.
7. 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 prvacy/confidentiality by removing your name and other direct identifiers, such as SSN or medical record number, from data shared with other laboratories.

New York residents only: By initialing this section, I confirm that I am a New York state resident, and I give permission for LMM to retain any remaining sample longer than 60 days after the completion of testing, and to be used as a de-identified sample for test development and improvement, internal validation, quality assurance, and training purposes. Otherwise, New York state law requires LMM to destroy my sample after 60 days, and it cannot be used for test development. Please initial here if you wish to give permission to maintain your isolated DNA: _____

RESEARCH POLICIES & OPPORTUNITIES

Blood or other samples sent to the 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 IRB approved 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 Laboratory for Molecular Medicine 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 in which you/your child may be able to participate.

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

ORDERING PROVIDER SIGNATURE

New York State residents excluded, require lab to obtain full informed consent

I, _____ (print name), as ordering provider, certify that the patient being tested and/or their legal guardian have been informed of the risks, benefits, 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.

_____

Signature (Ordering Provider) Date

Please Note: A patient consent form is available on our website (Partners.org/PersonalizedMedicine/Laboratory-For-Molecular-Medicine/Ordering/Policies) for your convenience and DOES NOT need to be returned to the LMM.

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APOL1 REQUISITION FORM

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

TEST TO BE PERFORMED

Please check box(es) to order.

❑ *APOL1* Genotyping (Ser342Gly, Ile384Met, & Asn388_Tyr389del) $400

CLINICAL INFORMATION

Clinical status: ❑ Affected ❑ Unknown ❑ Unaffected
Purpose of study: ❑ Diagnostic ❑ Risk Assessment ❑ Family history ❑ Other _____

ICD-10 Code(s): _____

Clinical Diagnosis: ❑ CKD ❑ ESKD ❑ FSGS ❑ HIV-nephropathy ❑ HTN-associated nephropathy
❑ Sickle cell nephropathy ❑ Other_____

Is this patient being considered as a living kidney donor? ❑ Yes ❑ No

Laboratory Values:
Creatinine levels Baseline_____ Current_____
Proteinuria ❑ Macro ❑ Micro ❑ None

Previous Genetic Testing: ❑ Yes ❑ No Gene(s)/Tests:_____
Result (if variants detected, please elaborate):_____

Has another family member already had genetic testing for this disease? ❑ Yes ❑ No
If yes, please describe and attach a copy of the genetic test lab report and pedigree.

FAMILY HISTORY

Family History: ❑ Yes ❑ No (Sketch below or attach pedigree if appropriate)

Paternal Ancestry:_____
Maternal Ancestry:_____
Consanguinity: ❑ Yes ❑ No

○ = Female □ = Male ◇ = Gender Unspecified
● ■ ◆ = Affected Individual ⊙ = Carrier

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APOL1 Genotyping Test Information

Background Information:

- The APOL1 gene contains two risk alleles, termed G1 and G2, which are associated with increased risk of non-diabetic nephropathy.
- Non-diabetic nephropathy is an umbrella term for a variety of kidney disease without diabetes mellitus as the underlying cause and can lead to end-stage renal disease.
- The G1 and G2 alleles are present at high frequency in the African American population. They are mutually exclusive and present on separate chromosomes.
    - G1: APOL1, NM_003661.3, c.[1024A>G;1152T>G], p.[(Ser342Gly;Ile384Met)]
        - This variant is present at a frequency of 21% in African Americans.
    - G2: APOL1, NM_003661.3, c.1164_1169del, p.(Asn388_Tyr389del)
        - This variant is present at a frequency of 13% in African Americans.
- African Americans have a significantly increased rate of non-diabetic nephropathy compared to individuals of other ancestries, even after adjusting for socioeconomic status, lifestyle, and other health factors.[1]
- APOL1 genotyping is an important consideration for kidney transplantation from living or deceased donors, as high-risk genotypes are associated with more rapid failure of transplanted kidneys and an increased risk for post-donation chronic kidney disease in living kidney donors.[2]

Inheritance Pattern:

- Risk of non-diabetic nephropathy due to the APOL1 risk alleles follows an autosomal recessive pattern.
- The presence of two risk alleles is associated with increased risk:
    - G1/G1
    - G1/G2
    - G2/G2
- Approximately 12% of African Americans have two APOL1 risk alleles and increased risk of kidney disease

What does this test include?

- This test determines the presence or absence of the G1 and G2 risk alleles in the APOL1 gene.
- This test is performed via Sanger sequencing of exon 6 of APOL1.

Who is this test indicated for?

- African Americans with kidney disease.
- African Americans being evaluated as a living kidney donor.

Test Outcomes:

- *Positive:*

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- The presence of two risk alleles (either G1/G1, G1/G2, or G2/G2) indicates an increased risk of non-diabetic nephropathy.

- *Carrier:*
  - The presence of one risk allele indicates that the individual is not at increased risk of non-diabetic nephropathy, but other family members could be carriers or at increased risk.
- *Negative:*
  - The presence of zero risk alleles indicates that the individual is NOT at increased risk of non-diabetic nephropathy. For individuals with a diagnosis of nephropathy or chronic kidney disease, alternate etiologies should be considered.

References:

1. Freedman BI, Limou S, Ma L, Kopp JB. APOL1-Associated Nephropathy: A Key Contributor to Racial Disparities in CKD. Am J Kidney Dis. 2018 Nov;72(5 Suppl 1):S8-S16. doi: 10.1053/j.ajkd.2018.06.020. PMID: 30343724; PMCID: PMC6200346.

2. Mena-Gutierrez AM, Reeves-Daniel AM, Jay CL, Freedman BI. Practical Considerations for APOL1 Genotyping in the Living Kidney Donor Evaluation. Transplantation. 2020 Jan;104(1):27-32. doi: 10.1097/TP.0000000000002933. PMID: 31449181; PMCID: PMC6933073.