PARTNERS HEALTHCARE | PERSONALIZED MEDICINE

The LMM is a satellite facility of Massachusetts General Hospital. • CLIA # 22D1005307

Laboratory for Molecular Medicine

65 Landsdowne Street • Cambridge, MA 02139
Phone: (617) 768-8500 • Fax: (617) 768-8513
www.partners.org/personalizedmedicine/lmm

SPECIMEN INFORMATION

Specimen: ❑ Blood ❑ DNA ❑ Other _____
❑ Cord Blood* ❑ CVS* ❑ Amnio*
❑ DNA* derived from:
(Choose One) ❑ Cord Blood ❑ CVS ❑ Amnio

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

Space for Lab Use Only

**Maternal specimen is required to perform Maternal Cell Contamination testing. For MCC sample, complete only the first page of the requisition form to submit with specimen. Note an additional $950 charge will be added for duplicate analysis, MCC studies, internal control tests and rush processing.*

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. 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 www.partners.org/personalizedmedicine/lmm.

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. 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 removing your name and other direct identifiers, such as SSN or medical record number, from data shared with other laboratories.

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 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. 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)

ORDERING PROVIDER SIGNATURE

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 (www.partners.org/personalizedmedicine/Laboratory-For-Molecular-Medicine/Ordering/Policies) for your convenience and DOES NOT need to be returned to the LMM.

CONNECTIVE TISSUE DISORDERS REQUISITION FORM

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

TEST(S) TO BE PERFORMED

Please check box(es) to order.

All panel tests are performed via next-generation sequencing (NGS). CNV analysis is included when NGS data meets necessary quality standards.

**MLPA of FBN1 is included when CNV via NGS data does not meet quality standards.*

Connective Tissue Disorders Tests

____ ❑ Connective Tissue Disorders Panel (14 genes)*

____ ❑ Familial Thoracic Aortic Aneurysms and Aortic Dissection (TAAD) Panel (9 genes)

❑ *REFLEX* to Connective Tissue Disorders Panel*

Familial Variant Testing (*Sequencing - first 3 variants, $500; each additional variant, $50.*)

____ ❑ Familial Variant(s) OR ❑ Research Confirmation - *If proband testing was performed elsewhere, please attach a copy of the original result and send positive control sample, if available.*

Gene_____Variant_____LMM Accession #: PM-_____

Proband Name_____Relationship to Patient_____

CLINICAL INFORMATION

Clinical Status: ❑ Affected ❑ Unknown ❑ Unaffected

Purpose of Study: ❑ Diagnostic ❑ Family history ❑ Prenatal ❑ Other _____

ICD-10 Code(s):
- ❑ Q87.40 (Marfan's syndrome, unspecified)
- ❑ Q87.410 (Marfan's syndrome with aortic dilation)
- ❑ Q87.418 (Marfan's syndrome with other cardiovascular manifestations)
- ❑ Q87.42 (Marfan's syndrome with ocular manifestations)
- ❑ Q87.43 (Marfan's syndrome with skeletal manifestations)
- ❑ Other _____

Suspected Diagnosis: *please check all that apply* **Age at diagnosis:** _____

❑ **Marfan syndrome** - *meets Ghent Criteria* ❑ Yes ❑ No ❑ **Loeys-Dietz syndrome** ❑ **Shprintzen-Goldberg syndrome**

❑ **Familial Thoracic Aortic Aneurysms & Dissections** ❑ **Ehlers-Danlos syndrome** - Type IV ❑ **Other**_____

CLINICAL FEATURES

Cardiovascular:

Aortic dilation ❑ Yes ❑ No
- ❑ Ascending
- ❑ Sinuses of valsalva

Z-score:_____

Aortic dissection ❑ Yes ❑ No
- ❑ Ascending
- ❑ Descending

❑ Mitral valve regurgitation
❑ Mitral valve prolapse
❑ Bicuspid aortic valve
❑ Arterial tortuosity
❑ Pulmonary hypertension
❑ Congential heart disease
Type: _____

Other:
❑ Organ rupture - Organ_____
❑ Seizures ❑ Strokes

Craniofacial
❑ High arch palate
❑ Cleft palate
❑ Micrognathia
❑ Retrognathia
❑ Hypertelorism
❑ Bifid uvula
❑ Other_____

Skin:
❑ Loose/stretchy skin
❑ Translucent skin
❑ Easy bruising
❑ Atrophic scaring
❑ Striae
❑ Livedo reticularis
❑ Hernia - Type:_____
❑ Other skin findings_____

Ocular findings:
❑ Myopia
❑ Cataracts
❑ Glaucoma
❑ Lens dislocation - Direction:_____
❑ Iris flocculi
❑ Blue/gray sclerae

Musculo-Skeletal:
❑ Hypermobility
❑ Scoliosis
❑ Joint pain
❑ Arachnodactyly
❑ Club foot
❑ Thumb sign
❑ Wrist sign
❑ Pectus
- ❑ Excavatum ❑ Carinatum

❑ Contractures
❑ Upper to lower segment ratio:_____
❑ Arm span to height ratio:_____

Additional Clinical Features:_____

_____

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

PLEASE PROCEED TO PAGE 4 TO COMPLETE THE FAMILY HISTORY

Laboratory for Molecular Medicine • 65 Landsdowne Street • Cambridge, MA, 02139
Phone: 617-768-8500 • Fax: 617-768-8513 • Website: www.partners.org/personalizedmedicine/lmm • Email: lmm@partners.org

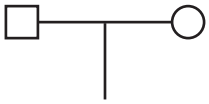
Last Revised: 22 Sep 2015

CONNECTIVE TISSUE DISORDERS REQUISITION FORM

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

FAMILY HISTORY

Family History: ❑ Yes ❑ No *If yes*, sketch below, attach pedigree, or list affected relatives.
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.



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

Laboratory for Molecular Medicine • 65 Landsdowne Street • Cambridge, MA, 02139
Phone: 617-768-8500 • Fax: 617-768-8513 • Website: www.partners.org/personalizedmedicine/lmm • Email: lmm@partners.org

Last Revised: 22 Sep 2015