


LABORATORY FOR MOLECULAR MEDICINE
 CLIA# 22D1005307
 Center for Genetics and Genomics
 65 Landsdowne Street
 Cambridge, MA 02139-4232
 Phone: (617) 768-8500
 Fax: (617) 768-8513
 The LMM is a satellite facility of Massachusetts General Hospital.



Patient Healthcare Card Information

Specimen type(s): _____ (MM-DD-YYYY)
 Blood DNA Other: _____ Date Collected: _____

REFERRING PHYSICIAN INFORMATION

First Name	MI	Last Name	Provider UPIN#	Speciality:
Phone:			Fax:	
Institution:			Contact if different from referring physician (e.g.genetic counselor) Please include name and phone/email:	
Address:			Fax duplicate report to (attach additional sheet if needed):	
City:	State:		DOB (MM-DD-YYYY):	
Zip Code:	Country:		Gender:	

PATIENT INFORMATION

First Name	MI	Last Name	Institution:
Phone:			Medical Record #:
Email:			Is the patient adopted? <input type="checkbox"/> Yes <input type="checkbox"/> No
Address:			Is patient deceased? <input type="checkbox"/> Yes <input type="checkbox"/> No
City:	State:		If yes, at what age?
Zip Code:	Country:		DOB (MM-DD-YYYY):
			Gender:

Race: (optional) American Indian or Alaska Native Asian
 Black or African American Caucasian
 Native Hawaiian or Other Pacific Islander
 (Check all that apply)

Ethnicity: (optional) Hispanic Yes No
 Ashkenazi Jewish Yes No
 Other: _____
 (Check all that apply)

PAYMENT INFORMATION

Referring Facility: _____
 Bill to name: _____ and/or Department: _____
 Facility address: _____
 Contact name: _____ Phone number: _____
 Purchase order number: _____

Patient Pay: Credit card Check (Test will not be performed until payment is received.)
 Name (as it appears on credit card): _____ Expiration Date: _____
 Credit card type: Mastercard Visa AMEX
 Credit card number: _____ 3 Digit Security Code: _____

How did you hear about our lab? Internet Site: _____ Colleague: _____
 Conference: _____ Other: _____

ALL TESTS ALSO REQUIRE TEST SPECIFIC FORM AND INFORMED CONSENT.

SAMPLE REQUIREMENTS:

The preferred blood specimen is a 7 ml blood sample (3-5ml for infants) collected in a lavender top (K3EDTA) blood tube. Smaller blood samples or other tissue specimens may also be acceptable for certain tests. Please contact the laboratory for more details.

SHIPPING SAMPLES:

Each sample must be accompanied by a requisition form (available at <http://www.hpcgg.org/LMM/forms.html>). Please ensure that all samples are labeled with the patient's name and date of birth. All genetic tests require informed consent. Please have the patient (or parent) read and sign the consent form 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 procedures, see our website <http://www.hpcgg.org/lmm/>.

INFORMED CONSENT:

I understand that:

- 1) The purpose of this test is to determine if I/my child may have a mutation in the gene(s) being tested, which has been found to be associated with this condition.
- 2) Genetic counseling is available to me if I desire further information about this condition.
- 3) Except in the case of a known mutation test, a negative genetic test result does not rule out a diagnosis of, a predisposition towards, or the ability to pass on this condition but diminishes the likelihood that this gene is involved.
- 4) This genetic test is specific for the indication for testing and does not test for other conditions. Therefore, a negative result does not guarantee my/my child's health.
- 5) In some families, genetic testing might discover non-paternity, or some other previously unknown information about family relationships, such as adoption.
- 6) The testing process includes highly skilled technicians and advanced technology. Although the method is extremely reliable, as in any laboratory, there is a small possibility that the test will not work properly, or an error may occur.
- 7) The lab will make every attempt to report results in the indicated turn-around time but cannot accept responsibility for delays.
- 8) If this test requisition form is incomplete, and my health care provider cannot provide the information, I understand that it may be necessary for lab staff to contact me directly to obtain or verify the information needed to complete the form.
- 9) I give permission to be contacted directly concerning research studies for the condition for which I am being tested. These studies may involve one or more of the following: 1) A request for additional clinical records about my condition, 2) Giving permission for my remaining sample to be included in studies looking for other causes or modifiers of this condition, 3) Learning about opportunities to be included in studies on newly developed treatments for my condition.

I do NOT give permission to be contacted directly about any research studies. _____ (Initial)

I have been informed and agree that my ordering provider will receive the results of my genetic tests and the ordering provider will discuss the results with me.

If my ordering provider is within **Partners HealthCare System**, I understand that my results will also become part of my permanent medical record. If this genetic test is for diagnostic purposes (condition which I am currently showing signs and symptoms) others may be able to view these test results as described in the **Partners HealthCare Notice for Use and Sharing of Protected Health Information** (http://intranet.partners.org/finance/hipaa/Privacy/1/1_PH122_EngVer.pdf). If this genetic test is for screening purposes (condition which I am not showing signs or symptoms) the test results will only be accessible within my record by my ordering provider.

I have carefully read and understand the above, have had any questions explained to my satisfaction, and do hereby consent to provide a specimen for testing.

 Name of patient (please print)

 Signature (Patient or patient's legal representative)

 Date

 Signature (Physician)

 Date

Please note: A physician may sign this form in lieu of the patient if prior consent has been obtained from the patient and if testing is for diagnostic purposes only.

MARFAN SYNDROME I/II and LOEYS-DIETZ SYNDROME
DISEASE SPECIFIC REQUISITION FORM

Name: _____

DOB: ____ / ____ / ____ (MM/DD/YYYY)

GENE TEST TO BE PERFORMED:

Check box to order test. Indicate order of testing in the space provided (i.e. 1, 2, 3).
Testing can be conducted concurrently if desired.

____ *FBN1* Sequencing

____ *TGF β R1* Sequencing

YES Check this box if you **DO WANT** the results of the *TGF β R1* *6A variant included on the report.

TGF β R1 *6A has been associated with cancer risk, but not with Marfan or Loeys-Dietz syndromes

____ *TGF β R2* Sequencing

Familial Known Mutation Test

Indicate gene and mutation here: _____

Comments/Special Instructions:

PURPOSE OF STUDY

Clinical status: Symptomatic Asymptomatic

Purpose of study: Diagnostic Carrier Screen Presymptomatic Prenatal

Other _____

Has another family member already had genetic testing for this disease? Yes No

If yes, please describe in the comments section and attach a copy of the genetic test lab report and pedigree.

Meets clinical criteria Yes No ICD-9 Code: 759.82 (Marfan) Other: _____

Known or Suspected Clinical Diagnosis: Marfan syndrome Loeys-Dietz syndrome

Age at Diagnosis: _____

Clinical Manifestations:

Skeletal findings:

Arachnodactyly Yes No

Scoliosis Yes No

Pectus deformity Yes No

Type: _____

High arched palate Yes No

Cardiovascular findings:

Aortic dilatation Yes No

Aortic dissection Yes No

Valve regurgitation Yes No

Type: _____

Valve prolapse Yes No

Type: _____

Ocular findings:

Lens dislocation Yes No

Myopia Yes No

Retinal detachment Yes No

Glaucoma Yes No

Cataracts Yes No

Dural ectasia Yes No

Pneumothorax Yes No

Other: _____

Surgery Yes No

Type: _____

Medications: _____

Upper to Lower Segment Ratio: _____

Arm Span to Height Ratio: _____

Facial features Yes No

Type: _____

Wrist sign: Yes No

Thumb sign: Yes No

Family history: Yes No List affected family members: _____

(Sketch or attach pedigree if appropriate)

Pedigree:

Standard Symbols:

○ = Female	■ ● ◆ = Affected individual
□ = Male	◉ = Carrier
◇ = Gender unknown	

Ethnicity

Paternal side: _____

Maternal side: _____

Consanguinity? Yes No

Other: _____

