

<p><b>LABORATORY FOR MOLECULAR MEDICINE</b></p> <p>CLIA# 22D1005307</p> <p>Center for Genetics and Genomics 65 Landsdowne Street Cambridge, MA 02139-4232 Phone: (617) 768-8500 Fax: (617) 768-8513</p> <p>The LMM is a satellite facility of Massachusetts General Hospital.</p>	<p>Patient Healthcare Card Information</p> 
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**Specimen type(s):**

Blood      Date Collected: \_\_\_\_\_ (MM-DD-YYYY)

**REFERRING PHYSICIAN INFORMATION**

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

**PATIENT INFORMATION**

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

<p><b>Race:</b> (optional)</p> <p><input type="checkbox"/> American Indian or Alaska Native      <input type="checkbox"/> Asian</p> <p><input type="checkbox"/> Black or African American                      <input type="checkbox"/> Caucasian</p> <p><input type="checkbox"/> Native Hawaiian or Other Pacific Islander</p> <p style="text-align: center;">(Check all that apply)</p>	<p><b>Ethnicity:</b> (optional)    Hispanic    <input type="checkbox"/> Yes    <input type="checkbox"/> No</p> <p>Ashkenazi Jewish    <input type="checkbox"/> Yes    <input type="checkbox"/> No</p> <p>Other: _____</p> <p style="text-align: center;">(Check all that apply)</p>
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**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](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.**

Name: \_\_\_\_\_ DOB: \_\_\_\_ / \_\_\_\_ / \_\_\_\_ (MM/DD/YYYY)

## GENE TEST TO BE PERFORMED:

## NOONAN SPECTRUM DISORDERS (NOONAN, LEOPARD, CARDIO-FACIO-CUTANEOUS, and COSTELLO SYNDROMES)

 Noonan Spectrum Gene Chip (PTPN11, SOS1, RAF1, KRAS, BRAF, MEK1, MEK2, HRAS)

Contact the laboratory for individual gene sequencing tests

## FAMILIAL KNOWN MUTATION TEST

 Familial Mutation Test (Indicate gene, mutation, and information on proband (1st person tested) below)

Gene \_\_\_\_\_ Mutation \_\_\_\_\_ LMM Accession #: PM- \_\_\_\_\_

Proband Name \_\_\_\_\_ Relationship To Proband \_\_\_\_\_

## PURPOSE OF STUDY

Clinical status:  Symptomatic  AsymptomaticPurpose 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.

Clinical diagnosis:  Noonan  LEOPARD  ICD-9 Codes:  759.89 (Noonan/LEOPARD)  
(check all known/suspected clinical diagnoses)  CFC  Costello  759.8 (Other specified abnormalities)Age at diagnosis: \_\_\_\_\_  Other \_\_\_\_\_Congenital heart defect:  Pulmonic valve stenosis  Hypertrophic cardiomyopathy  None Septal defect Other \_\_\_\_\_

Facial dysmorphism:

 Epicanthal folds Ptosis of the eyelids Low nasal bridge Hypertelorism Downward eye slant Low set ears and posteriorly rotated Papillomas Coarseness None

Short stature:

 Yes - Height(%): \_\_\_\_\_ Parental Heights: \_\_\_\_\_ No

Cognitive development:

 Learning disabilities Developmental delay Mental retardation Normal

Skeletal:

 Pectus excavatum Pectus carinatum Scoliosis Normal

Genitourinary:

 Cryptorchidism (undescended testes) Normal Kidney malformation

If yes, please describe: \_\_\_\_\_

Bleeding diathesis:

 Yes No

If yes, please describe: \_\_\_\_\_

Hair/Skin findings:

 Yes No

If yes, please describe: \_\_\_\_\_

Malignancy:

 Yes No

If yes, please describe: \_\_\_\_\_

Other: \_\_\_\_\_

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

## Comments/Special Instructions:

## Pedigree:

## Standard Symbols

○ = Female

□ = Male

◇ = Gender unknown

⊙ = Carrier

■ ● ◆ = Affected individual

Ethnicity Paternal side: \_\_\_\_\_

Maternal side: \_\_\_\_\_

Consanguinity?  Yes  No

Other: \_\_\_\_\_