

**LABORATORY FOR MOLECULAR MEDICINE**

CLIA# 22D1005307

Center for Genetics and Genomics

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**PARTNERS**  
HEALTHCAREHARVARD  
MEDICAL SCHOOL

The LMM is a satellite facility of Massachusetts General Hospital.

**Specimen type(s):**

(MM-DD-YYYY)

 Blood DNA Other: \_\_\_\_\_

Date Collected: \_\_\_\_\_

**REFERRING PHYSICIAN INFORMATION**

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

**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
Address:		If yes, at what age?		
		DOB (MM-DD-YYYY):	Gender:	
City:	State:	_____ - _____ - _____	<input type="checkbox"/> Male <input type="checkbox"/> Female	
Zip Code:	Country:		<input type="checkbox"/> Unknown	

<b>Race:</b> (optional)	<input type="checkbox"/> American Indian or Alaska Native	<input type="checkbox"/> Asian	<b>Ethnicity:</b> (optional) Hispanic <input type="checkbox"/> Yes <input type="checkbox"/> No	
	<input type="checkbox"/> Black or African American	<input type="checkbox"/> Caucasian		Ashkenazi Jewish <input type="checkbox"/> Yes <input type="checkbox"/> No
	<input type="checkbox"/> Native Hawaiian or Other Pacific Islander			Other: _____
(Check all that apply)			(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 A SPECIFIC GENE TEST 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.**

# REQUISITION FORM

FAMILIAL ADENOMATOUS POLYPOSIS (FAP) AND HEREDITARY NON-POLYPOSIS COLORECTAL CANCER (HNPCC)

Name: \_\_\_\_\_

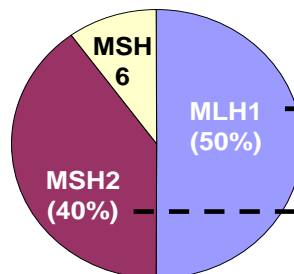
DOB: \_\_\_\_ / \_\_\_\_ / \_\_\_\_ (MM/DD/YYYY)

## HNPCC GENE TESTS

Reflex ?  Yes  No  
(If yes, indicate order below)

- ↓
- \_\_\_\_\_  *MLH1* Full Gene Sequencing
  - \_\_\_\_\_  *MSH2* Full Gene Sequencing
  - \_\_\_\_\_  *MSH6* Full Gene Sequencing
  - \_\_\_\_\_  *MLH1* Deletion/Duplication (MLPA)
  - \_\_\_\_\_  *MSH2* Deletion/Duplication (MLPA)

Mutation Spectrum  
(Point Mut + Del/Dup)



Mutation Detection Rate

- 50-80% SEQU  
5-10% DEL/DUP
- 90-95 % SEQU  
17-50 % DEL/DUP

### HNPCC Familial Known Mutation Test

Gene \_\_\_\_\_ Proband's Accession# \_\_\_\_\_  
Mutation \_\_\_\_\_ Patient's Relationship to Proband \_\_\_\_\_

Peltomaki et al (2003); www.genetests.org

## FAP GENE TESTS

Reflex ?  Yes  No   
(If yes, indicate order below)

- ↓
- \_\_\_\_\_  *APC* Full Gene Sequencing (~ 90% of *APC* mutations)
  - \_\_\_\_\_  *APC* Deletion/Duplication (MLPA) (~ 10% of *APC* mutations)

### APC Familial Known Mutation Test

Mutation \_\_\_\_\_ Proband's Accession# \_\_\_\_\_  
Patient's Relationship to Proband \_\_\_\_\_

## PURPOSE OF STUDY

**Clinical Diagnosis**  HNPCC  FAP  Attenuated FAP  Unaffected  
 Gardner syndrome  Turcot Syndrome  Muir Torre Syndrome  Unknown

Age at Diagnosis: \_\_\_\_\_ ICD-9 Code(s): \_\_\_\_\_

Meets Clinical Diagnostic Criteria  Yes  No

If yes, which criteria are met? \_\_\_\_\_

Microsatellite Instability (MSI) analysis performed?

- Yes  No
- MSI High  MSI low  MSI Stable

Immunohistochemistry (IHC) performed?  Yes  No

- MLH1* negative  *MSH6* negative
- MSH2* negative  *PMS2* negative

### Clinical Manifestations:

*Colon polyps*  Yes  No #: \_\_\_\_\_ *Colon cancer*  Yes  No Age of onset: \_\_\_\_\_

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

Family history:  Yes  No List affected family members, their clinical features, and relationship to this patient here or on the attached form:

\_\_\_\_\_  
\_\_\_\_\_

**Pedigree:**

**Standard Symbols:**

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

**Ethnicity**

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

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

**Consanguinity?**     Yes     No

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