

HNPCC Familial Mutation Testing INSTRUCTIONS FOR SAMPLE SUBMISSION

Documentation: Each sample must be accompanied by:

1. A requisition for DNA analysis completed by the physician, nurse or genetic counselor requesting screening. **Please note: ICD-9 code is required for billing purposes. If ICD-9 code is unknown, please provide patient's clinical symptom(s) or family history that prompted testing.**
2. The patient's pedigree to include three generations, if possible.
3. An informed consent signed by the patient (if under 18 years of age, the parent or guardian should sign) and the professional obtaining the consent. Please have the patient initial at the top of each page and send **all** pages of the consent.
4. A verification of blood tubes form signed by the patient, parent or guardian. The form should be signed at the time of the blood draw.
5. A completed registration form with check, money order, credit card authorization or information for billing the referring institution.

IN THE EVENT THAT ALL PROPERLY COMPLETED FORMS DO NOT ACCOMPANY THE SPECIMEN, YOU WILL BE NOTIFIED, AND TESTING WILL BE HELD UNTIL PAPERWORK IS COMPLETE.

Preparing Sample:

- Obtain 2 EDTA tubes (lavender top) of blood - approx. 4 mL per tube
- Label each tube with the patient's name and date sample was obtained
- We accept banked or recently extracted DNA; please include the concentration.
- For prenatal testing: cultured amniotic fluid or CVS cells, 2 confluent T-25 flasks. Please call the lab prior to sending a prenatal sample. We are often able to offer testing on a direct villi or amnio sample, and we can discuss the requirements with you. 5mL of whole blood from each parent should accompany the prenatal sample.

Shipping Sample: Ship at room temperature via Federal Express or other overnight courier that guarantees AM delivery to arrive Monday-Friday. There is no one in the laboratory evenings and weekends to receive samples. If sample is drawn on a Friday, please refrigerate it until shipment on the following business day.

Shipping Address: Genetic Diagnostic Laboratory
University of Pennsylvania
415 Anatomy-Chemistry Building
3620 Hamilton Walk
Philadelphia, PA 19104

**GENETIC DIAGNOSTIC LABORATORY
UNIVERSITY OF PENNSYLVANIA SCHOOL OF MEDICINE
DEPARTMENT OF GENETICS
Tel: (215) 573-9161 • Fax: (215) 573-5940
CLIA ID: 39D0893887**

REQUEST FOR HNPCC FAMILIAL MUTATION TESTING

PATIENT FIRST NAME _____ PATIENT LAST NAME _____

BIRTH DATE _____ SEX _____ RACE _____ ETHNICITY _____

STREET ADDRESS _____

CITY _____ STATE _____ ZIP _____ HOME PHONE _____

Please include a three generation pedigree.

Who in the family has had genetic testing for HNPCC and where was testing performed?

What was the result? _____

Does the patient have clinical symptoms associated with HNPCC? No Yes

REFERRING PHYSICIAN _____ PHONE _____ FAX _____

GENETIC COUNSELOR _____ PHONE _____ FAX _____

EMAIL ADDRESS FOR COUNSELOR OR PHYSICIAN _____

INSTITUTION and

DEPARTMENT _____

STREET ADDRESS _____

CITY _____ STATE _____ ZIP _____ COUNTRY _____

ICD-9 CODE (patient's clinical symptoms) _____

TEST REQUESTED

Analysis for known familial sequencing mutation hMLH1 hMSH2 hMSH6

Prenatal diagnosis of known familial sequencing mutation hMLH1 hMSH2 hMSH6

Please include a copy of genetic result for affected family member
For all prenatal requests, please call the laboratory to discuss requirements; 5mL of whole blood from both
parents should accompany any prenatal sample for MCC studies.

Turnaround time for a familial mutation is 2-3 weeks and for prenatal diagnosis is 1-3 weeks.

CPT Codes: Familial mutation-83891, 83898x6, 83904x6, 83912
Prenatal diagnosis-83891x2, 83898x4, 83904x5, 83912

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Patient Registration Form

Please provide the following information. We cannot perform your test without ALL of this information. PLEASE PRINT ALL ANSWERS

PATIENT INFORMATION

LAST FIRST MI

PATINET'S MAIDEN NAME IF PREVIOUS TESTING WAS PERFORMED

Date of Birth: _____

Gender: ___ Male ___ Female

Patient Address: _____
STREET/APT NO.

CITY, STATE, ZIP COUNTRY IF OUTSIDE UNITED STATES

HOME TELEPHONE

REFERRING PHYSICIAN

NAME MD DO OTHER

INSTITUTION

STREET ADDRESS

CITY, STATE, ZIP

TELEPHONE

FAX

PAYMENT OPTIONS (must choose one) [a receipt will be mailed to patient for self-pay options]

_____ I have enclosed a check payable to the "Genetic Diagnostic Laboratory" for \$ _____

_____ Please charge my credit card for the amount of \$ _____
___ VISA ___ Master Card ___ Discover ___ American Express

Card Number: _____ Exp date: _____

Name as it appears on card: _____

_____ I have Pennsylvania Medicaid. A copy of my Medicaid card is attached.

_____ INSTITUTIONAL BILLING: The Institution where my testing originated has agreed to pay all charges for the testing. INCLUDE Billing Address, Person Authorizing Payment, Telephone, and Fax below:

BILLING ADDRESS

BILLING ADDRESS

PERSON AUTHORIZING PAYMENT

PHONE

FAX

Verification of Correctly Identified Blood Tubes

I am a participant in genetic DNA testing.

I have been shown the tubes containing my blood for this genetic testing and my name has been correctly placed on each one of these tubes.

I have signed a copy of the consent form regarding this genetic testing to be sent along with my blood samples. I have been given a copy of the consent form to keep.

Participant Name: _____

Participant/Parent Signature: _____

Date: _____

Initials _____

FAMILIAL MUTATION

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**Informed Consent: Genetic Testing for a Known Familial Mutation in a
Non-Polyposis Colorectal Cancer (HNPCC) Gene**

BACKGROUND:

Most colon cancer is not inherited, but in 5-10% of cases, the cancer may be due to an alteration in a gene inherited from a parent. When individuals from families that meet the criteria for HNPCC are tested, approximately 70% are shown to have an alteration in one of the four genes identified so far. Two of the four different genes that have been identified as causing HNPCC (MSH2 and MLH1) are responsible for a large majority of the alterations. Additional mutations have been found in MSH6.

PURPOSE:

The purpose of this genetic testing is to determine whether I, or my child, have inherited the altered HNPCC gene that has been identified in my family and is known to be involved in the development of colon cancer which runs in families. I understand the testing will take approximately 1-3 weeks to complete. I have provided the Genetic Diagnostic Laboratory of the University of Pennsylvania with the specifics of that mutation. Individuals who inherit an altered HNPCC gene are at greatly increased risk of colorectal cancer when compared to individuals who do not have an altered gene. Both men and women with an alteration in an HNPCC gene are at increased risk of developing colorectal cancer. Alterations in HNPCC genes may also be associated with an increased risk for other cancers including uterine, ovarian, stomach and urinary tract. I have had the opportunity to discuss the benefits and drawbacks of HNPCC testing for myself. I understand that there are limitations as to what these test results can tell me. This testing is intended to provide me with the most accurate estimate possible about my lifetime chance of developing colon cancer and whether I may be at increased risk for other cancers. I understand that this test will not provide me with any information about the current status of my health.

TESTING PROCEDURE:

Genetic testing requires several teaspoons (2 tubes) of blood. Before my blood is drawn, I will watch as my name, or my child's name, is written correctly on empty blood tubes and after my blood is drawn I will sign a form indicating that I have positively identified the tubes containing my blood. The blood sent to the laboratory will be divided into two separate samples. The first sample will be used to complete the genetic testing. The second sample will be tested to confirm the diagnosis if the first sample indicates an alteration. There will be no additional charge for the confirmation testing. No information pertaining to me will be provided to any of my relatives without my consent. No information will be provided to me by phone or mail from the University of Pennsylvania, regardless of the outcome.

If this is a prenatal sample, cells from either a CVS or amniocentesis will be used to isolate fetal DNA for analysis of the DNA mutation found in my family that causes colon cancer. In order to confirm that analysis was performed on fetal cells, studies to rule out any maternal cell contamination will also be performed.

RESULTS:

When results of the laboratory testing are available, I will be given the option of postponing or declining disclosure of these results. If I choose to learn the results, I will be given this information as part of a counseling session. I understand that there are two possible results to this testing:

1. I may learn that I, or my child or my fetus, have the altered HNPCC gene which has been identified in my family. If the altered gene is present, there is an increased risk for developing colon cancer.

2. I may learn that the testing did not detect the altered HNPCC gene.

RISKS AND DISCOMFORTS:

I understand that there is usually a minimal amount of risk involved in drawing a blood sample. These include pain at the blood drawing site, bleeding, bruising and infection.

The risks of disclosure of information regarding my genetic susceptibility to colorectal cancer include depression, anxiety, anger, and fear of the future. This result could affect my relationships with family members and loved ones. I understand that if I learn that I have an altered HNPCC gene my ability to obtain health, disability or life insurance could be affected. Certain health, disability and life insurance companies may consider an inherited HNPCC gene alteration to be a "pre-existing condition," and I may be responsible for disclosing this information prior to obtaining new health or life insurance.

Some individuals may experience feelings of guilt or other forms of anxiety if they are found not to have an altered gene when other family members did inherit an altered gene.

If this is a prenatal sample, I have been counseled on the risks of the prenatal procedure, and I understand that the genetic analysis of the HNPCC DNA mutation found in my family will determine whether my fetus has inherited the altered gene and for the future risk to develop colon cancer.

BENEFITS OF RECEIVING INFORMATION:

This study may provide information that will make it possible to determine more precisely whether I or my children are at high risk of developing colorectal cancer or other cancers. My results will not be released to any relative or any other third party without my express written consent.

Providing me with the most accurate information currently available regarding my inherited cancer risk may enable me to make better choices about my health care needs. If I do have an altered gene, this information may allow me to make decisions about colorectal cancer screening or preventative surgery that may decrease my risk of developing fatal complications from the cancer.

If I have inherited the altered HNPCC gene, each of my children has a 50% risk of inheriting the mutation. Genetic counseling is available for at-risk family members to discuss the benefits of genetic testing. The option for prenatal diagnosis is available for future pregnancies.

If I have not inherited the HNPCC gene alteration, I cannot pass this on to any children, and I may experience some sense of relief as a result.

LIMITATIONS OF DNA TESTING FOR HNPCC:

The identification of an alteration in one of the HNPCC genes does not mean that a cancer will definitely develop. It only means there is a high risk for cancer to develop. We cannot predict when cancer may develop. Continual cancer screening and consultation with a gastroenterologist is recommended.

USE OF SPECIMENS:

I understand that any blood or tissue specimens obtained for the purposes of this genetic testing become the exclusive property of the Genetic Diagnostic Laboratory. After the specific tests requested have been completed and reported, the Laboratory may dispose of, retain, or preserve these specimens and may use these specimens for research. I understand that my or my child's identity will be protected and that research results will not be provided to me or to any other party. If there are new developments in the field, my physician/genetic counselor may be contacted by the Genetic Diagnostic Laboratory staff to offer me the opportunity to have additional clinical testing.

ALTERNATIVE TO GENETIC TESTING:

I understand that my participation in this testing is completely voluntary and will not affect my medical treatment now or in the future. The alternative is not to undergo testing; in which case I will not learn whether I have an altered form of an HNPCC gene. This decision is perfectly acceptable.

REQUEST FOR MORE INFORMATION: I understand that I may ask more questions about this testing and my results at any time. At the Genetic Diagnostic Laboratory, Susan Walther, MS (215-573-9161) or Arupa Ganguly, PhD, FACMG (215-898-3122) will be available to answer questions as they arise. I will be given a copy of this consent form to keep.

I have explained to _____ the purpose of this genetic testing, the procedures required and the possible risks and benefits to the best of my ability.

Printed Name of Professional Obtaining Consent

Signature of Professional Obtaining Consent

Date

CONSENT OF PATIENT:

I have read and received a copy of this consent form. I agree to have genetic testing and accept the risks. I understand the information provided in this document, and I have had the opportunity to ask questions I might have about the testing, the procedure, the associate risks and the alternatives.

Printed Name of Patient

Patient's DOB

Signature of Patient

Date

CONSENT OF PARENT OR GUARDIAN:

I have read and received a copy of this consent form. I agree to have genetic testing performed for my child and accept the risks. I understand the information provided in this document, and I have had the opportunity to ask questions I might have about the testing, the procedure, the associate risks and the alternatives.

Printed Name of Parent/Guardian

Relationship to Child

Signature of Parent/Guardian

Child's Name

Child's DOB