**REQUEST FOR LYNCH SYNDROME FAMILIAL MUTATION TESTING**

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

### PATIENT INFORMATION*

<table>
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<tr>
<th>FIRST NAME</th>
<th>MI</th>
<th>LAST NAME</th>
<th>BIRTH DATE (MM/DD/YYYY)</th>
<th>GENDER</th>
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**ANCESTRY**
- [ ] Western/Northern European
- [ ] Central/Eastern European
- [ ] Latin American/Caribbean
- [ ] African
- [ ] Asian
- [ ] Jewish (Ashkenazi)
- [ ] American Indian
- [ ] Near East/Middle Eastern
- [ ] Native Hawaiian or Pacific Islander

Specify countries: ________________________________________________________________  [ ] Other: ____________________

### CLINICAL INFORMATION

**ICD-10 CODE(S):***
- [ ] C18.9 Malignant tumor of colon unspecified
- [ ] C26.0 Malignant neoplasm of intestinal tract, unspecified
- [ ] K63.5 Polyp of colon
- [ ] Other: ___________________________________________________________________

[ ] C54.1 Malignant neoplasm of endometrium
- [ ] Z80.0 Family history of malignant neoplasm of gastrointestinal tract
- [ ] Z84.81 Family history of genetic disease carrier

Personal history of cancer? [ ] No  [ ] Yes; if yes, describe: ____________________________________________

Family history of cancer? [ ] No  [ ] Yes; if yes, describe:

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<tr>
<th>RELATIONSHIP</th>
<th>CANCER DIAGNOSIS</th>
<th>AGE DIAGNOSED</th>
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Laboratory only offers FAMILIAL ANALYSIS for a KNOWN MUTATION:

Name of person previously tested and relationship: ___________________________________________

Was the previous testing performed at the Genetic Diagnostic Laboratory? [ ] Yes  [ ] No

Result (Please include a copy of the result):*  ____________________________________________

### TEST REQUESTED

- [ ] Analysis of known familial sequencing mutation: hMLH1
- [ ] Analysis of known familial sequencing mutation: hMSH2
- [ ] Analysis of known familial sequencing mutation: hMSH6
- [ ] Prenatal analysis of known familial sequencing mutation: hMLH1**
- [ ] Prenatal analysis of known familial sequencing mutation: hMSH2**
- [ ] Prenatal analysis of known familial sequencing mutation: hMSH6**

* Required information
** Please call the laboratory prior to sending a prenatal sample. Please refer to the special requirements for prenatal samples on the Instructions for Sample Submission page.

9/28/2015
PATIENT REGISTRATION FORM

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

PATIENT INFORMATION

FIRST NAME                      MI                      LAST NAME                      BIRTH DATE (MM/DD/YYYY)                      GENDER

STREET ADDRESS

CITY                                                STATE                                                ZIP

PHONE

PHYSICIAN INFORMATION*

REFERRING PHYSICIAN                      PHONE                      FAX

GENETIC COUNSELOR                      PHONE                      FAX

EMAIL ADDRESS FOR COUNSELOR                                                EMAIL ADDRESS FOR PHYSICIAN

INSTITUTION AND DEPARTMENT

STREET ADDRESS

CITY                                                STATE                                                ZIP

PAYMENT OPTIONS* (must choose one) [a receipt will be mailed to the 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 of cardholder 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

NAME OF INDIVIDUAL 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: __________________
INFORMED CONSENT: GENETIC TESTING FOR A KNOWN FAMILIAL MUTATION IN GENE RELATED TO LYNCH SYNDROME

Background: Lynch syndrome (also known as hereditary nonpolyposis colorectal cancer) is related to variations in MLH1, MSH2, MSH6, PMS2 and the EPCAM genes. Mutations in these genes increase the risk to develop many cancers including colorectal, uterine, gastrointestinal tract, urinary tract, and ovaries. Lynch syndrome is inherited in an autosomal dominant pattern, meaning that first degree relatives have a 50% chances of inheriting the familial mutation. Not all people who inherit the mutation will develop cancer.

Purpose: The diagnostic samples will be used for the purpose of attempting to determine if I (or my child) am/is a carrier of an altered TP53 gene related to a hereditary predisposition for cancer. This information may help establish appropriate medical management.

Results: I understand that there are five possible results to this testing:

PATHOGENIC VARIANT: A clinically significant variant is detected in one of the genes analyzed. This may explain personal or family history of cancer. My or my child’s healthcare provider will make medical management recommendations based on this information.

LIKELY PATHOGENIC VARIANT: A variant is detected in one of the genes analyzed which is the likely deleterious. This may explain personal or family history of cancer. My or my child’s healthcare provider will make medical management recommendations based on this information.

VARIANT OF UNCERTAIN SIGNIFICANCE: The laboratory may detect an alteration in the genes analyzed which is currently of unknown significance, called a “variant of unknown significance (VUS)”. The laboratory will work with your physician to help determine if the VUS can be further classified as to whether it is disease-causing for Lynch syndrome.

LIKELY BENIGN VARIANT: A variant is detected in one of the genes analyzed which is not likely to be clinically significant. This result reduces the likelihood that I, or my child, have a mutation in the genes tested.

NEGATIVE: No clinically significant mutations were identified in the genes analyzed. This result reduces the likelihood that I, or my child, have a mutation in the genes tested. Methods currently in use are unable to detect all mutations and therefore may still carry a DNA mutation that was not detected by the current technology.

Disclosure Policy: Results will be reported to me only through the physician or genetic counselor who requested the testing due to the complexity of DNA based testing. The results of genetic testing are protected by the Health Insurance Portability and Accountability Act (HIPAA) of 1996 (Public Law 104-191). Release of test results is limited to authorized personnel as required by law. Additionally, results can be released to other medical professionals or other parties with my written consent.

Limitations: While genetic testing is highly accurate for detection of the majority of disease causing mutations, a small fraction of mutations may be missed by the current technology. Due to the nature of the testing, there is a small possibility that the test will not work properly or that an error will occur. Occasionally, testing may reveal a variant of unknown significance that is unable to be definitively interpreted as positive or negative for disease-association based on our current knowledge of the variant. My signature below acknowledges my voluntary participation in this test, but in no way releases the laboratory and staff from their professional and ethical responsibility to me. Furthermore, the DNA analysis performed at the University of Pennsylvania Genetic Diagnostic Laboratory is specific only for MLH1, MSH2 or MSH6 analysis, and in no way guarantees my health.

There are federal laws in place that prohibit health insurers and employers from discriminating based on genetic information (i.e. Genetic Information Nondiscrimination Act (GINA) of 2008 (Public Law 110-233). There are currently no laws specific to discrimination based on genetic information for life insurance, long term care, or disability insurance companies. Additional information about GINA can be found on the Genetics Public & Policy Center’s website at www.dnpolicy.org.

9/28/2015
Use of Specimens: 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 for research or for validation in the development of future genetic tests. In all circumstance described previously, my identity will be protected and research results will not be provided to me or to any other party. If use of this genetic material results in a scientific publication, it will not contain any identifying information. Indicate consent or denial to the above sentence by initialing below. My refusal to consent to research will not affect the reporting of my genetic results.

_____ I consent to the use of my DNA sample for future test validation and/or research purposes.
_____ I do not consent to the use of my DNA sample for future test validation or research purposes.

In the event that my sample is used for research purposes, the laboratory may wish to contact my physician/genetic counselor for additional information regarding my sample. This includes, but is not limited to, information on personal health and family history as it relates to the genetic testing. 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. Indicate consent or denial to the above sentence by initialing below. My refusal to consent to research will not affect the reporting of my genetic results.

_____ I consent to be contacted by the Laboratory in the future for research purposes.
_____ I do not consent to be contacted by the Laboratory in the future for research purposes.

Genetic Counseling provided by a qualified specialist (i.e. genetic counselor/medical geneticist) is a recommendation for individuals proceeding with genetic testing. This service is available before and after genetic testing. Additionally, other testing or further physician consults may be warranted. The Genetic Diagnostic Laboratory is also an available resource to ask more questions about this testing. The laboratory genetic counselor can be reached at 215-573-9161 and Arupa Ganguly, PhD, FACMG can be reached at 215-898-3122. I will be given a copy of this consent form to keep.

HEALTHCARE PROVIDER STATEMENT:

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 performed for myself, or my fetus, and accept the risks. I understand the information provided in this document and I have had the opportunity to ask questions I have about the testing, the procedure, the associate risks and the alternatives.

Patient’s Printed Name: ___________________________   DOB: ________________

Patient’s Signature: ___________________________   Date: ________________
(or Parent/Guardian if patient is a minor)
Name and Relationship: ___________________________
(Parent/Guardian if patient is a minor)

Initials _______