Hartford Hospital
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Consent for Hereditary Cancer Genetic Testing for APC and MYH

Introduction. This form describes the benefits, risks, and limitations of genetic testing for inherited susceptibility to cancer. Please read this form carefully and discuss any questions that you have with your team of providers.

Purpose. This test analyzes a specific gene or gene(s) for genetic changes called mutations. The gene(s) analyzed are associated with a specific hereditary cancer syndrome. This test will help determine if a person has a significantly increased risk of developing certain tumors due to a mutation(s) in a cancer-predisposing gene. Not everyone who has a gene mutation will develop cancer.

Adenomatous polyposis syndromes are hereditary cancer syndromes which are associated with multiple adenomatous polyps. Individuals who carry an *APC* gene mutation may have either Familial Adenomatous Polyposis (FAP) or Attenuated Familial Adenomatous Polyposis (AFAP). A mutation in both copies of the *MYH* gene is associated with MYH-associated Adenomatous Polyposis (MAP). These persons have a greater risk to develop colorectal cancer than those who do not have a mutation.

The cancer risks associated with *APC* or *MYH* gene mutation(s) are listed below. The percentages cited are estimates and may change as more research is done.

FAP: early onset of adenomas (average: 15 years of age and 75% of individuals by age 20); early onset colorectal cancer (average 35 years of age); if untreated, development of cancer is inevitable. The mean age of colon cancer diagnosis in untreated individuals is 39 years (range 34-43 years). FAP is also associated with duodenal / ampullary carcinoma, thyroid cancer, childhood hepatoblastoma, gastric carcinoma, rare CNS tumors, upper GI polyps, cutaneous soft tissue tumors, osteomas, supernumerary teeth, congenital hypertrophy of the retinal pigment epithelium, desmoid tumors, and mesenteric fibromatosis.

AFAP is characterized by a significant risk for colon cancer but fewer colonic polyps (average of 30), more proximally located polyps, and diagnosis of colon cancer at a later age; management may be substantially different.

MAP: People with MAP often develop multiple adenomatous polyps and colonic and extracolonic cancers. *MYH* carriers have an increased risk of breast cancer.

Both men and women carrying a mutation within the *APC* gene have a 50% (1 in 2) chance of passing it on to each of their children. Each child of an individual carrying one *MYH* gene mutation would have a 25% (1 in 4) chance to be affected with MAP if that individual's partner also carried one *MYH* gene mutation. For an individual affected with MAP, the chance of each of his/her children being affected with MAP would be 50% (1 in 2) if their partner carries a mutation in one copy of their two *MYH* genes.

Test Procedure. *Hartford Hospital* personnel will draw approximately 2 tubes of blood and send it to the appropriate genetic testing laboratory. The laboratory will analyze your *APC* and/or *MYH* genes to look for mutations. No other tests will be run on your sample without your written consent.

You are consenting to the following tests:

- ❒ Comprehensive testing: *APC* sequencing and rearrangement and *MYH mutation panel*
- ❒ MYH sequencing and rearrangement analysis
- ❒ Single Site analysis

_____Patient initials

Test Results and Interpretation. Listed below are the possible results from testing:

- Positive – A mutation that is associated with an increased risk for hereditary cancer was identified. Knowing this information may help you and your doctor make more informed choices about your health care, such as screening, risk-reducing surgeries and preventive medication strategies.
- Negative – A mutation was not identified.
    - If you are the first person tested in your family, you still have at least the same risk of cancer as does a person in the general population. You may still be at greater than average risk for hereditary cancer due to a genetic predisposition that cannot be detected by this test, either in the gene(s) you were tested for or in another gene linked to hereditary cancer.
    - If you test negative for a mutation known to be in your family, you are considered to have the same risks as others in the general population.
- Uncertain – A genetic change (a Variant of Uncertain Significance, or VUS) was detected but it is not known if this change is linked to cancer risk. You still have at least the same risk of cancer as the general population, and may still be at greater than average risk due to a genetic predisposition that cannot be detected by this test.

The genetic counselor will discuss the results with you and what they mean. Genetic tests results have implications for blood relatives. If you choose, the genetic counselor will help you inform family members if a mutation is found.

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Benefits. Your genetic test results may help you and your doctor make more informed choices about your health care, such as screening, risk-reducing surgeries and preventive medication strategies. The identification of gene mutation(s) in a family enables other blood relatives to determine whether or not they share the same hereditary cancer risks. If you test positive, you should discuss with your healthcare provider how hereditary cancer is inherited and learn about the chance your children and blood relatives may have inherited the same mutation(s) in the gene(s) tested. If you test negative for a known mutation in your family, you cannot pass on that mutation to your children. If you have had cancer or a pre-cancerous condition, you would still have some increased risk based on your own history. If you have not had cancer, you would still have the general population risk of cancer.

Risks. Learning that you have a genetic mutation may be upsetting and may increase feelings of depression, anxiety, and vulnerability. You could feel that you have to make difficult decisions about your medical care. Learning of a positive result may affect relationships with family members. News of an inconclusive result may increase feelings of distress. If more than one family member is tested, there is a possibility of learning sensitive information about your family, such as non-paternity or undisclosed adoption.

To address concerns regarding possible health insurance discrimination, most states and the federal government have enacted laws to prohibit genetic discrimination. Furthermore, broad federal legislation (HIPAA) prohibits unauthorized disclosure of confidential personal health information. However, for the most part, these laws do not prevent life and disability insurers from using genetic testing information in determining coverage. **It is our policy to include genetic information and test results into your medical record.**

Limitations. This test analyzes only the *APC* and/or *MYH* genes. If no mutation is found, you may still be at risk for hereditary cancer due to a genetic predisposition that cannot be detected by this test, either in the gene(s) you were tested for or in another gene linked to hereditary cancer. Genetic testing clarifies cancer risks for only those cancers related to the genes analyzed.

Financial Responsibility. Genetic testing of appropriate individuals is typically reimbursed by health insurance. You are responsible for any cost of the genetic test not reimbursed by insurance, including all applicable co-pays and deductibles.

Future Correspondence. Due to the dynamics of this field, there continues to be new information and data. It is recommended that you keep in contact with your healthcare provider, annually, to learn of any new developments in cancer genetics and to provide any updates to your personal or family history which may affect your cancer risks.

I confirm that the possible benefits, risks, costs and limitations of genetic testing for *APC* and *MYH* have been explained to me. All my questions have been answered. I have read this consent form and will be given a copy for my records. My signature below indicates my willingness to have genetic testing for the *APC* and/or *MYH* gene mutation(s). I understand the confidentiality policy outlined in the consent form. This means that I recognize that genetic information and a copy of the test result will be included in my medical record.

_____ _____ _____
Signature of Patient Date Time

_____
Print name

I have explained the possible risks, limitations and costs as well as potential benefits of genetic testing for mutations in *APC* and/or *MYH* and have answered any questions regarding the test to the best of my ability.

_____ _____ _____
Genetic Counselor/Clinician Date Time

_____Patient initials