Patient Summary
Genetic Testing for Lynch Syndrome for People with a New Diagnosis of Colorectal Cancer

Recommendations from the EGAPP™ Working Group

The independent, non-federal Evaluation of Genomic Applications in Practice and Prevention (EGAPP™) Working Group (www.egappreviews.org/workingrp.htm) reviewed the scientific evidence for genetic testing for Lynch syndrome (hereditary colorectal cancer) and developed a recommendation statement about the appropriate use of this testing. This brief summary of the EGAPP™ recommendation statement can help the general public understand what is intended by this recommendation and where to find more information.

This information may be helpful for people with a recent diagnosis of colorectal cancer (cancer of the colon or rectum) and their close family members.

Disclaimer: Our office does not conduct research on individual genetic conditions and does not offer medical advice to individuals. If you have specific concerns about your health, we suggest that you discuss them with your doctor.

What is the purpose of genetic testing for Lynch syndrome for people newly diagnosed with colorectal cancer?
Genetic testing is used to find out if a person’s colorectal cancer is hereditary (caused by an inherited gene change), so that family members can learn if they are also at increased risk. This could help to protect them from getting this disease.

What is Lynch syndrome?
About 3% of colorectal cancer cases are due to an inherited condition known as Lynch syndrome (sometimes referred to as hereditary nonpolyposis colorectal cancer or HNPCC). People with this condition have a greatly increased chance to develop colorectal cancer, especially at a young age (younger than 50 years). Children, sisters, and brothers of people with Lynch syndrome have a 50% chance to inherit the condition. Parents and other blood relatives such as grandparents, aunts, uncles, nieces and nephews are also at increased risk to have Lynch syndrome.

Who developed these recommendations?
The EGAPP™ Working Group is a group of scientists and health care experts who review available research and evidence to make recommendations about the use of genetic tests. This independent, non-government body includes representatives from universities, industry, clinical practice, insurance companies, and public health.

Did EGAPP™ recommend the use of genetic testing for newly diagnosed colorectal cancer patients?
• YES: The EGAPP™ Working Group found good scientific evidence to show that if individuals with colorectal cancer are found to have Lynch syndrome by genetic testing, their family members can benefit by:
  o Undergoing genetic testing to learn if they are also at increased genetic risk.
  o If positive for the gene change, having earlier and more frequent screening which can prevent colorectal cancer.
• They concluded that all people with a new diagnosis of colorectal cancer should be offered counseling and educational materials about genetic testing for Lynch syndrome.

Other Information
Are there other people the test might help?
Although the EGAPP™ recommendation did not address use of testing in other situations, people with colorectal cancer diagnosed in the past (especially under age 50), and/or people with several family members with colorectal and/or uterine cancer may also benefit from genetic evaluation for Lynch syndrome.

How do I find out more about the condition/test?
In addition to talking with your health care provider, the Web sites below provide additional information on Lynch syndrome, cancer genetic testing, and access to genetic counseling services.
• Colorectal Cancer, Centers for Disease Control and Prevention (www.cdc.gov/cancer/colorectal/)
• Cancer Genetic Services Directory, National Cancer Institute (www.cancer.gov/search/geneticsservices/)
• Find-A-Counselor, National Society of Genetic Counselors (www.nsgc.org/resourcelink.cfm)