American College of Medical Genetics **ACT**

Family History ACT Sheet

Colon Cancer (Asymptomatic)

Condition Description: Colon cancer is preceded by an abnormal growth of cells that initially form a polyp which if not removed can grow into cancer. Although most colon cancer occurs without a positive family history there are a significant number of families that have an increased familial risk and a smaller number with inherited risk due to mutations in single genes. These inherited forms include hereditary nonpolyposis colon cancer-Lynch Syndrome (HNPCC) (~3%), and familial adenomatous polyposis (FAP) or adenomatous polyposis coli due to mutations in the APC gene (<1%). Removal of the precancerous polyp allows prevention of colon cancer.

Family History: A current family history, to include at least grandparents, parents, aunts and uncles, siblings and children is an important means of identifying individuals at high risk. The usual family history is most accurate for close relatives (1st degree relatives – parents, children) and less accurate for more distant relatives (grandparents, aunts, uncles). Affected first degree relative(s) confer the highest risk. Key issues that identify inherited cancer include earlier age of onset (<50 years), organ of origin of cancer(s), and affected individuals in multiple generations.

ACTIONS YOU SHOULD TAKE TO IDENTIFY HIGH RISK INDIVIDUALS:

- Take or update the family history for cancer and intestinal polyps.
 - Age of onset and primary location of cancer.
 - Information on noncolorectal cancers.
- Ask and facilitate the family's ability to obtain confirmation of cancer diagnosis by pathology report, in each affected individual with focus on all first degree relatives – including histologic characteristics and immunohistochemistry where available.
- Obtain and review results of any cancer genetic testing has been done.
- Refer to cancer genetics specialist if appropriate.
- Colonoscopy is the single most effective means of preventing colon cancer.

Clinical Considerations: Colon cancer remains the 3rd leading cause of death due to cancer. The two common types of inherited colon cancer are HNPCC and FAP. In general, the younger the patient is at the time colorectal cancer is diagnosed the more likely it is to be genetic. Risk indicators include three or more family members with HNPCC-related cancers, affected individuals in two successive generations, one or more of the HNPCC-related cancers diagnosed prior to age 50. Other tumors found in people with HNPCC include endometrial cancer, ovarian cancer, stomach cancer and others. FAP usually presents with multiple polyps, and it is important to recognize since virtually all individuals with it will eventually develop colon cancer. Colonoscopy in individuals with a family history of full FAP (that is thousands of polyps) should begin at age 10 years. Individuals from families with attenuated FAP (associated with tens to a hundred polyps) or HNPCC should begin colonoscopy at least 10 years before the earliest cancer in the family. Management of individuals at high risk of colon cancer includes genetic testing, repeated endoscopic screening, and prophylactic surgery.

Additional Information:

National Cancer Institute
FAP
HNPCC
ClinGen Actionability Report

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that heroome available after that date.



American College of Medical Genetics ACT

OCAL RESOURCES: Insert State newborn screening program web site links	
State Resource site (insert state newborn screening program website information)	
Name	
URL	
Comments	
Local Resource Si	te (insert local and regional newborn screening website information)
Name	
URL	
Comments	
DDENIDIV. Deserves	s with Full LIDL Addresses

APPENDIX: Resources with Full URL Addresses

Additional Information:

National Cancer Institute

http://www.nci.nih.gov/cancertopics/types/colon-and-rectal

FAP

 $\underline{http://ghr.nlm.nih.gov/condition=familial adenomatous polyposis}$

HNPCC

http://ghr.nlm.nih.gov/condition=hereditarynonpolyposiscolorectalcancer

ClinGen Actionability Report

https://actionability.clinicalgenome.org/ac/Adult/ui/stg2SummaryRpt?doc=AC069

Referral (local, state, regional and national):

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

