

Clinical Cancer Genetics Clinical Referral Guidelines

Gastrointestinal (GI) Center 2011 Update

Strongly encourage referral:

Patients with any of the following:

- Prior tumor studies suggestive of Lynch syndrome (MSI-H and/or loss of staining for any mismatch repair protein by IHC)
- Colorectal cancer diagnosed <50 years
- Colorectal cancer diagnosed at any age AND first- or second-degree relative with any HNPCC-related cancers*, diagnosed <50 years
- Colorectal cancer, regardless of age AND one or more of the following (Lynch syndrome-associated cancers due to mutations in MLH1, MSH2, MSH6, or PMS) in his/her personal history:
 - Synchronous or metachronous colorectal cancer
 - Endometrial cancer
 - Sebaceous neoplasias
 - Gastric, pancreas, ureter and renal pelvis, biliary tract, brain, or small intestinal cancers
- Multiple (> 10) adenomas, on a single colonoscopy
- Hamartomatous polyps, any number, occurring at any age (manifestations of Peutz-Jeghers syndrome, juvenile polyposis syndrome)
- Diffuse gastric adenocarcinoma (linitis plastica) AND first- or second-degree relative with gastric cancer or lobular breast cancer (manifestations of hereditary diffuse gastric cancer due to mutations in CDH1)
- Pancreas carcinoma, regardless of age, AND of Ashkenazi Jewish ancestry
- Pancreas carcinoma AND personal or family history of pancreatic cancer
- Pancreas cancer AND personal history, or first- or second- degree relative with breast cancer < 50 and/or ovarian cancer
- Family history of a known mutation for a cancer predisposition syndrome

Consider referral:

Patients with any of the following:

- Colorectal cancer diagnosed >60 years AND first- or second-degree relative with any HNPCC-related cancers*, regardless of age
- Multiple (>5) adenomas on a single colonoscopy, under age 50
- Unusual polyp burden (young age at diagnosis, histology, number)
- No personal history of cancer, but a family member who meets any of the above (Strongly encourage referral) criteria
- Pancreas carcinoma and personal or family history of melanoma diagnosed under age 40 years

* HNPCC-related cancers include: colorectal, endometrial, ovarian, gastric, pancreas, ureter and renal pelvis, biliary tract, brain, small intestinal cancers and sebaceous gland adenomas and keratoacanthomas (revised Bethesda guidelines, Umar *et al*, JNCI 2004)