

2014 Practice Bulletins

#147 ACOG Practice Bulletin, November: Lynch Syndrome

This is an autosomal dominant, inherited syndrome primarily involving (particularly in young aged women) cancer of the colon and rectum, but also cancer of ovary and endometrium. In addition, it is also associated with small bowel, stomach, hepatobiliary, renal and ureteric cancers. Appropriate screening for these patients allows for decreased morbidity and mortality associated with these conditions. The general prevalence is about 1 in 1500 and it accounts for about 5% of inherited uterine cancers and 10% of inherited ovarian cancers. This syndrome is characterized by an inability to repair individual DNA base pair errors.

Guidelines for those with endometrial or colorectal cancer, and who may have concomitant

Lynch Syndrome:

- Patients are at increased risk for Lynch Syndrome if they developed:
 - ✓ Endometrial or colorectal cancer diagnosed prior to age 50
 - ✓ Endometrial or ovarian cancer with other Lynch or Hereditary Non Polyposis Colon Cancer (HNPCC), at any age
 - ✓ Colorectal cancer containing lymphocytes, mucinous or signet-ring cells, diagnosed before age 60
 - ✓ Colorectal or endometrial cancer plus a 1st degree relative with Lynch / Hereditary Non Polyposis Colon Cancer that was diagnosed prior to age 50
 - ✓ Endometrial or colorectal cancer diagnosed at any age with 2 or more 2nd degree or closer relatives, with Lynch/Hereditary Non Polyposis Colon Cancer, diagnosed at any age
- Lynch syndrome testing detects either actual tumor DNA, or microsatellite (mismatched pair) instability. These tests are:
 - ✓ Immunochemistry
 - ✓ Microsatellite Instability testing
 - ✓ MLH1 promoter methylation testing
- Patients diagnosed with endometrial or colon cancer should be tested where possible, for

Lynch syndrome by any 1 of the following 3 options:

- 1) If she is deemed to be 'at risk' by the guidelines (mentioned above), then one should test the tumor tissue (using the tests mentioned above)
- 2) Test tumor tissue of all endometrial or colorectal cancers regardless of age of diagnosis
- 3) Test tumor tissue of all endometrial or colorectal cancers, if cancer occurred prior to age 60 (this preferred by Society of Gynecological Oncology)

Should any women without cancer be tested for Lynch syndrome?