**Evidence-based Practice Guidelines on Genetic Susceptibility Testing for**

**Lynch Syndrome**

**Identifying Colorectal Cancer Patients at Risk for Lynch Syndrome:**

**Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Recommendation** (2009)1

* The EGAPP Working Group found sufficient evidence to recommend offering screening or genetic testing for Lynch syndrome (LS) *to all individuals with newly diagnosed colorectal cancer*

**Identifying Patients at Risk for Lynch Syndrome Who Do Not Have Colorectal Cancer**

* National Comprehensive Cancer Network (NCCN) (2014) Recommendations2
  + Referral for genetic counseling for Lynch syndrome is recommended for:
    - Women with endometrial cancer diagnosed before age 50
    - Individuals in families with known Lynch syndrome
  + Amsterdam Criteria should be used to identify patients at risk due to family health history:
    - At least three relatives diagnosed with a cancer associated with Lynch syndrome (colorectal, endometrial, small bowel, ureter, renal-pelvis) and
      * One must be a first-degree relative of the other two
      * At least 2 successive generations must be affected
      * At least one relative with cancer associated with Lynch syndrome should be diagnosed before age 50
      * Familial Adenomatous Polyposis (FAP) should be excluded in the colorectal cancer cases and reported family health history should be verified when possible
* American College of Medical Genetics (ACMG) and National Society of Genetic Counselors (NSGC) Recommendations (2015)3
  + Referral for genetic counseling for Lynch syndrome is recommended if any of the following are present in a personal or family health history:
    - Colorectal or endometrial cancer diagnosed before age 50
    - Colorectal or endometrial cancer diagnosed at 50 or older and a first-degree relative with colorectal or endometrial cancer at any age
    - Synchronous or metachronous colorectal or endometrial cancers in the same person
    - Sebaceous adenoma or carcinoma and one or more additional case of any Lynch syndrome associated cancer\*\*
    - Colorectal cancer showing mismatch repair deficiency on tumor screening
    - 3 or more family members with Lynch syndrome associated cancers\*\*

\*\*Lynch syndrome associated cancers include: colorectal cancer, endometrial (uterine) cancer, urothelial cancers (ureter and renal collecting ducts), gastric cancer, ovarian cancer, small bowel cancer, glioblastoma, sebaceous adenocarcinoma, biliary tract cancer, and pancreatic cancer