HEREDITARY BREAST-OVARIAN CANCER SYNDROME

**Affected:** An individual with any of the following:
- Mutation identified in family member*
- Breast cancer diagnosed ≤ 45y
- Breast cancer diagnosed ≤ 50y with:
  - An additional breast cancer primary
  - ≥ 1 close relative¹ with breast cancer
  - ≥ 1 close relative¹ with pancreatic cancer
  - ≥ 1 close relative¹ with prostate cancer (Gleason score ≥7)
  - An unknown or limited family history
- Triple negative breast cancer diagnosed ≤ 60y
- Breast cancer diagnosed at any age with:
  - ≥ 1 close relative¹ with breast cancer diagnosed ≤ 50y
  - ≥ 1 close relative¹ with ovarian cancer at any age
  - A close relative¹ with male breast cancer
  - ≥ 2 close relatives¹ with breast cancer, pancreatic cancer, or prostate cancer (Gleason score ≥7) at any age
- Male breast cancer at any age
- Primary ovarian cancer (epithelial, non-mucinous) at any age
- Prostate cancer (Gleason score ≥7) or pancreatic cancer at any age with ≥1 close relative¹ with ovarian cancer at any age or breast cancer ≤50y or two relatives with breast, pancreatic, or prostate cancer at any age
- BRCA 1/2 mutation detected by tumor profiling in any tumor type

**Unaffected (whenever possible, testing should begin in an affected family member):**
An individual with either a maternal or paternal family history of any of the following:
- Mutation identified in family member*
- ≥ 3 breast primaries from the same side of the family; especially if one is diagnosed ≤ 50y
- Breast cancer diagnosed ≤ 45y
- Ovarian cancer diagnosed at any age
- Male breast cancer
- Member of an at risk population (i.e. Ashkenazi Jewish) with a family history of breast and/or ovarian cancer
- First- or second-degree relative meeting any of the criteria listed to the left

**REFER A PATIENT:** To refer a patient for genetic testing, please contact Justine Cooper, Markey genetic counselor, at 859-323-3083.
HEREDITARY COLON CANCER SYNDROMES

Lynch syndrome

- Mutation identified in family member*
- Individual meeting Amsterdam II Criteria
  - ³3 relatives with a cancer associated with LS²; all of the following criteria should be present
    - One relative must be first-degree relative of the other two
    - At least two successive generations affected
    - At least one relative’s cancer should be diagnosed before age 50y
- Individual meeting revised Bethesda Guidelines
  - Colon cancer diagnosed <50y
  - Synchronous or metachronous colorectal or other LS-related tumors², regardless of age
  - Colon cancer in a patient with ≥1 first-degree relatives with an LS-related cancer², with one of the cancers diagnosed before age 50y

Hereditary Polyposis

- Mutation identified in family member*
- Clinical diagnosis of FAP or other polyposis syndrome
- ≥ 10 adenomas in the same individual
- Individual with multiple gastrointestinal hamartomatous or hyperplasic polyps

GENERAL

- ≥ 3 close relatives¹ with a diagnosis of the same cancer type (≥2 relatives if the patient is one of the affected individuals)
- A single individual with multiple primary cancer diagnoses; especially if one was ≤ 50
- A combination of breast cancer and any one of the following cancers on the same side of the family:
  - Thyroid
  - Sarcoma
  - Adrenocortical carcinoma
  - Endometrial
  - Pancreatic
  - Brain
  - Diffuse gastric
  - Leukemia/Lymphoma
  - Dermatologic manifestations of Cowden Syndrome

¹Close relatives include first-, second- and third-degree relatives

² LS-related cancers include colorectal, endometrial, gastric, ovarian, pancreas, ureter and renal pelvis, biliary tract, brain (usually glioblastoma), and small intestinal cancers, as well as sebaceous gland adenomas and keratoacanthomas (as seen in Muir-Torre)

* A copy of the family member's test results is needed to test the patient

If you have a patient who you suspect may have a hereditary cancer syndrome but does not fit into these referral guidelines, please contact Justine Cooper (genetic counselor) at 859-323-3083 or at Justine.Cooper@uky.edu to discuss whether a referral for genetic counseling is appropriate.