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PATIENT EVALUATION

Patient referred to Clinic

Provider to assess need for Genetics referral, see Appendix A for Referral Criteria

Does patient meet primary referral criteria?

Refer patient to Genetics

Does patient meet additional referral criteria?

Genetic Counselor (GC) to assess:
- Patient’s relevant personal and family history
- Cancer risk/hereditary cancer risk
- Testing guidelines

Testing indicated?

Yes

- Consider Genetics referral
- Clinic to manage care as clinically indicated

No

Clinic to manage care as clinically indicated

Does patient agree to testing?

Yes

See Page 3 for testing and follow-up

No

Clinic to manage care as clinically indicated

RECOMMENDATION

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## Genetic Counseling

### Disclaimer:
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### Testing

- Patient signs lab- and test- specific genetic testing informed consent
- GC coordinates appropriate test(s)\(^1\) and reviews results(s)

### Result

#### Mutation positive
- GC discusses results and interpretation with patient and documents in the patient’s electronic health record
- Utilize National Comprehensive Cancer Network (NCCN) Clinical Practice Guidelines or other published recommendations
- Refer to the appropriate high risk clinic(s) based on the genetic mutation
- Consider referral to specialist to discuss family planning implications, as needed
- Encourage patient to discuss results with family members (for potential testing)\(^2\)

#### Mutation negative
- GC discusses results and interpretation with patient and documents in the patient’s electronic health record
- Service Center to manage care as clinically indicated
- Manage based on family history and refer to the appropriate high risk clinic(s) as necessary

#### Variant of uncertain significance (VUS)
- GC discusses results and interpretation with patient and documents in the patient’s electronic health record
- GC contacts patient for further discussion if an amended report of the VUS is received from the reference lab
- Manage based on family history and refer to the appropriate high risk clinic(s) as necessary

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1. In most cases peripheral blood is the preferred sample; in select cases (e.g., allogeneic stem cell transplant or hematologic malignancy) a different source of DNA such as cultured fibroblasts from a skin punch biopsy is required.

2. Refer to Appendix B for Patient Education Material.

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Approved by the Executive Committee of the Medical Staff on 10/15/2019
### APPENDIX A: Genetics Counseling Referral Criteria

<table>
<thead>
<tr>
<th>Primary Referral Criteria</th>
<th>Additional Referral Criteria</th>
</tr>
</thead>
<tbody>
<tr>
<td>● Patient with a personal history of breast cancer diagnosed at ≤ 50 years of age</td>
<td>Patients that do not meet Primary Referral Criteria, but have a personal history of breast cancer and there is a strong clinical suspicion for hereditary cancer (i.e., strong family history of early onset pancreatic cancer, prostate cancer, or melanoma)</td>
</tr>
<tr>
<td>● Patient with a personal history of TRIPLE NEGATIVE breast cancer diagnosed at ≤ 60 years of age</td>
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</tr>
<tr>
<td>● Patient with two breast primaries when first breast cancer is diagnosed ≤ 50 years of age</td>
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<tr>
<td>● Patient with a personal history of breast cancer diagnosed at any age, and one or more of the following:</td>
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<tr>
<td>○ Personal history of ovarian cancer or pancreatic cancer</td>
<td></td>
</tr>
<tr>
<td>○ Family history of ovarian cancer</td>
<td></td>
</tr>
<tr>
<td>○ Family history of breast cancer diagnosed at ≤ 50 years of age</td>
<td></td>
</tr>
<tr>
<td>○ Family history of male breast cancer</td>
<td></td>
</tr>
<tr>
<td>○ Family history of ≥ 2 relatives diagnosed with breast cancer at any age</td>
<td></td>
</tr>
<tr>
<td>○ Family history of metastatic or high grade (Gleason score ≥ 7) prostate cancer</td>
<td></td>
</tr>
<tr>
<td>○ Family history of pancreatic cancer</td>
<td></td>
</tr>
<tr>
<td>○ Family history of thyroid cancer, endometrial cancer, and/or dermatologic manifestations of Cowden syndrome</td>
<td></td>
</tr>
<tr>
<td>○ Family history of sarcoma, adrenocortical cancer, brain tumors, leukemia or lymphoma</td>
<td></td>
</tr>
<tr>
<td>○ Ashkenazi Jewish ancestry</td>
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</tr>
<tr>
<td>● Any male patient with a personal history of breast cancer</td>
<td></td>
</tr>
<tr>
<td>● Any member of a family with a known mutation</td>
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</tr>
<tr>
<td>● Metastatic breast cancer patient considering targeted therapy based on genetic test results (i.e., PARP inhibitors)</td>
<td></td>
</tr>
<tr>
<td>● Patient with BRCA1/2 pathogenic or likely pathogenic variant detected on tumor profiling on any tumor type in absence of germline pathogenic/likely pathogenic variant analysis</td>
<td></td>
</tr>
</tbody>
</table>

1 Family history should be all on the same side of the family (i.e., either maternal or paternal) and includes first, second, and third-degree relatives.
**APPENDIX A: Genetics Counseling Referral Criteria - continued**

<table>
<thead>
<tr>
<th>Gastrointestinal</th>
<th>Primary Referral Criteria</th>
<th>Additional Referral Criteria</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Patients with any of the following:</td>
<td>Patients with any of the following:</td>
</tr>
<tr>
<td></td>
<td>● Prior tumor studies suggestive of hereditary nonpolyposis colorectal cancer (HNPCC) syndrome (MSI-high and/or loss of staining for any mismatch repair protein by IHC), regardless of tumor type</td>
<td>● Colorectal cancer diagnosed at any age and first- or second-degree relative with any HNPCC-related cancer¹</td>
</tr>
<tr>
<td></td>
<td>○ If loss of MLH1/PMS2, no evidence of MLH1 methylation and/or no somatic BRAF mutation (in primary colorectal tumors)</td>
<td>○ Multiple (&gt; 5) adenomas on a single colonoscopy at &lt; 50 years of age</td>
</tr>
<tr>
<td></td>
<td>● Colorectal adenocarcinoma diagnosed at &lt; 50 years of age</td>
<td>● Unusual polyp burden (young age at diagnosis, histology, number)</td>
</tr>
<tr>
<td></td>
<td>● Colorectal adenocarcinoma diagnosed at any age and first- or second-degree relative with any HNPCC-related cancers¹, diagnosed at &lt; 50 years of age</td>
<td>¹ HNPPC-related cancers include: colorectal, endometrial, ovarian, gastric, pancreas, ureter and renal pelvis, biliary tract, brain, small intestinal cancers and sebaceous gland adenomas and keratoacanthomas (per revised Bethesda guidelines, Umar et al, JNCI 2004)</td>
</tr>
<tr>
<td></td>
<td>● Colorectal adenocarcinoma, regardless of age and one or more of the following in his/her personal history:</td>
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<tr>
<td></td>
<td>○ Synchronous or metachronous colorectal cancer</td>
<td></td>
</tr>
<tr>
<td></td>
<td>○ HNPCC-related cancers¹</td>
<td></td>
</tr>
<tr>
<td></td>
<td>● Multiple (&gt; 10) adenomas on a single colonoscopy or &gt; 20 lifetime cumulative adenomas</td>
<td></td>
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<tr>
<td></td>
<td>● Hamartomatous polyps, any number, occurring at any age</td>
<td></td>
</tr>
<tr>
<td></td>
<td>● Diffuse gastric adenocarcinoma (limitis plastica) diagnosed at or under 40 years of age</td>
<td></td>
</tr>
<tr>
<td></td>
<td>● Diffuse gastric adenocarcinoma (limitis plastica) regardless of age and a first- or second-degree relative with gastric cancer or lobular breast cancer.</td>
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</tr>
<tr>
<td></td>
<td>● Pancreatic adenocarcinoma</td>
<td></td>
</tr>
<tr>
<td></td>
<td>● Family history of a known mutation for a cancer predisposition syndrome</td>
<td></td>
</tr>
<tr>
<td></td>
<td>● Somatic test results concerning for a germline mutation</td>
<td></td>
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</tbody>
</table>

Continued on next page
### APPENDIX A: Genetics Counseling Referral Criteria - continued

<table>
<thead>
<tr>
<th>Gynecologic</th>
<th>Primary Referral Criteria</th>
<th>Additional Referral Criteria</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Patients with any of the following:</td>
<td>Patients with any of the following:</td>
</tr>
<tr>
<td></td>
<td>• High grade non-mucinous epithelial ovarian cancer, including primary peritoneal cancer and fallopian tube cancer</td>
<td>• Do not meet Primary Referral Criteria, but have a significant family history of cancer</td>
</tr>
<tr>
<td></td>
<td>• Endometrial cancer, and one or more of the following:</td>
<td>• Patient diagnosed with endometrial cancer at &lt; 50 years of age may be considered for referral at the clinician’s discretion particularly if known endometrial cancer risk factors (e.g., obesity) are absent</td>
</tr>
<tr>
<td></td>
<td>○ Personal history of colorectal cancer, regardless of age</td>
<td>• Endometrial cancer plus personal or family history of follicular thyroid cancer, breast cancer, and/or dermatologic manifestations of Cowden syndrome</td>
</tr>
<tr>
<td></td>
<td>○ First-degree relative with colorectal or endometrial cancer at any age</td>
<td></td>
</tr>
<tr>
<td></td>
<td>○ Any family history of colorectal or endometrial cancer diagnosed at &lt; 50 years of age</td>
<td></td>
</tr>
<tr>
<td></td>
<td>○ Microsatellite instability (MSI)/immunohistochemistry (IHC) suggestive of Lynch syndrome</td>
<td></td>
</tr>
<tr>
<td></td>
<td>• Family history of a known mutation for a cancer predisposition syndrome</td>
<td></td>
</tr>
</tbody>
</table>

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APPENDIX B: Patient Education Material

Hereditary Breast and Ovarian Cancer Syndrome

Lynch Syndrome: Hereditary Nonpolyposis Colorectal Cancer Syndrome (HNPCC)
https://www.mdanderson.org/patient-education/Genetics/Lynch-Syndrome-(HNPCC)_docx(pe).pdf

Cancer Genetics Overview

Genetic Counseling

Genetic Discrimination Laws

Family History: Gathering Information About Cancer

Familial Adenomatous Polyposis (FAP)
**SUGGESTED READINGS**


Genetic Counseling

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