Common types of gynecologic cancers include cervical, ovarian, endometrial (uterine), vaginal, and vulvar cancers. Patients may undergo germline and/or somatic testing at the time of diagnosis to inform treatment planning. Such testing may be called “molecular, genomic, genetic, or gene” testing.

According to clinical practice guidelines by the American Society of Clinical Oncology (ASCO), germline testing for BRCA1/2 and other ovarian cancer susceptibility genes for all women diagnosed with epithelial ovarian cancer is recommended.1 Germline test results may reveal an underlying hereditary cancer syndrome and/or inform therapy decisions (eg, PARP inhibitors for patients with a germline BRCA pathogenic variant). Some companion diagnostic tests use multiple methods to look for homologous recombination deficiency (HRD) and may identify patients with HRD-positive ovarian cancer who are eligible for certain PARP inhibitors.2 Somatic biomarker tests may also identify a genomic alteration (eg, an NTRK fusion) that would make the patient eligible for targeted therapy.

The following tips have been developed to support greater understanding of biomarkers in gynecologic tumors for the multidisciplinary cancer care team.

**BACKGROUND**

**HEREDITARY (GERMLINE) TESTING:** genetic testing that may reveal a hereditary cancer syndrome or may identify patients who may be eligible for certain types of targeted therapy.

**BIOMARKER OR TUMOR (SOMATIC) TESTING:** often performed using a multigene panel and/or other tests (eg, protein expression) to identify patients who may be eligible for targeted therapy and/or immunotherapy. Testing is usually performed on tumor tissue or blood.

**TIP**

**Clearly Differentiate Germline vs. Somatic Test Results**

In many cancer programs, test reports received from external reference labs are often scanned into an EHR “media” tab. As such, germline test reports may get mixed in with somatic test reports. Clearly differentiating which test reports are germline vs somatic is important to avoid confusion and better guide members of the treatment team.

**TIP**

**Establish Universal Reflex Testing for Endometrial Cancer**

Universal testing for MSI (microsatellite instability) or MMR (DNA mismatch repair) should be performed for all patients with endometrial cancer. These results may identify patients with Lynch Syndrome. Patients with MSI-High tumors may also be eligible for treatment with immunotherapy. The NCCN Guidelines® for Uterine Neoplasms recommend that HER2 testing also be performed for certain patients with advanced or metastatic disease.4

**TIP**

**Incorporate Biomarker Testing for Advanced Cervical Cancer**

The NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Cervical Cancer recommend PD-L1 and MSI/MMR testing for patients with recurrent, progressive, or metastatic cervical cancer.5 They also recommend NTRK gene fusion testing for patients with cervical sarcoma. For patients with advanced cervical cancer, the NCCN Guidelines® suggests considering comprehensive molecular profiling as determined by an FDA-approved assay, or a validated test performed in a CLIA-certified laboratory and RET gene fusion testing for patients with locally advanced or metastatic cervical cancer.

The following tips have been developed to support greater understanding of biomarkers in gynecologic tumors for the multidisciplinary cancer care team.
Patients may not know which types of testing they have received, especially when they do not have access to their test results. Some patients may refuse germline testing if they think they have already received it (e.g., they were told their tumor has undergone genomic profiling). Therefore, it is essential for patients to be educated about the types of tests that may appear on their pathology report (e.g., MSI/MMR result may be on the pathology report), and which types of tests will be released as a separate report (e.g., a hereditary genetic test report from an external reference lab).

It is also helpful to provide patients with written educational materials about their test results. These handouts should clearly indicate whether the patient received germline testing, somatic testing, or both. Ensure these handouts are available in multiple languages. Since patients are often overwhelmed at their first visit, it is important to follow up at subsequent visits and to ensure that patients understand how their test results will guide their care.

Some patient portals offer access to these results, so health systems should ensure that test reports clearly indicate which reference lab performed each test.

The National Comprehensive Cancer Network® (NCCN®) offers the NCCN Guidelines for Patients® for Ovarian Cancer in several languages and explains how different biomarker tests are used to guide treatment decisions. Since genetic counselors can help interpret germline and somatic test reports, it is important to include genetic counselors as members of the multidisciplinary cancer care team. Their input may help to identify more patients who are eligible for targeted therapy, especially when uncommon variants are identified. Genetic counselors may also identify situations where somatic test results may indicate that germline testing is also warranted.

Some patients may be reluctant to see a genetic counselor if it requires traveling long distances. Therefore, exploring whether telehealth options are available may enable more patients to receive genetic counseling. Telehealth may also make it easier to inform family members when cascade testing is indicated.

Germline and/or somatic tests are commonly ordered by gynecologic oncologists, medical oncologists, or genetic counselors, and interpreted and reported by pathologists. Identifying a multidisciplinary group of physician champions who can lead efforts to standardize testing processes, implement reflex testing protocols when appropriate, and track testing rates as quality improvement measures can be helpful.

Some cancer programs may do well communicating positive test results to patients but may identify communication of negative test results as an area of continuous improvement. Other programs may need to strengthen how they coordinate the communication of test results between pathology and oncologists, especially when reports are scanned into a generic media tab in the EHR or faxed to the outpatient provider.
REFERENCES:
2. Referenced with permission from the NCCN Guidelines for Patients® for Ovarian Cancer. V.2023. National Comprehensive Cancer Network, Inc. 2023. All rights reserved. Accessed July 10, 2023. To view the most recent and complete version of the guideline, go online to NCCN.org. NCCN makes no warranties of any kind whatsoever regarding their content, use, or application and disclaims any responsibility for their application or use in any way.
4. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Uterine Neoplasms. V.1.2024. National Comprehensive Cancer Network, Inc. 2024. All rights reserved. Accessed January 17, 2024. To view the most recent and complete version of the guideline, go online to NCCN.org. NCCN makes no warranties of any kind whatsoever regarding their content, use, or application and disclaims any responsibility for their application or use in any way.
5. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Cervical Cancer. V.1.2024. National Comprehensive Cancer Network, Inc. 2024. All rights reserved. Accessed January 17, 2024. To view the most recent and complete version of the guideline, go online to NCCN.org. NCCN makes no warranties of any kind whatsoever regarding their content, use, or application and disclaims any responsibility for their application or use in any way.
8. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic. V.3.2024. National Comprehensive Cancer Network, Inc. 2024. All rights reserved. Accessed February 13, 2024. To view the most recent and complete version of the guideline, go online to NCCN.org. NCCN makes no warranties of any kind whatsoever regarding their content, use, or application and disclaims any responsibility for their application or use in any way.
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