Guideline Support for Hereditary Breast and Ovarian Cancer Genetics

Clinical genetic testing for gene mutations allows more precise identification of those women who are at an increased risk of inherited breast cancer and ovarian cancer.

Evaluation of the likelihood of a patient having a gynecologic cancer predisposition syndrome enables physicians to provide individualized assessments of cancer risk.

1. ACOG Practice Bulletin Number 182, September 2017

ASPIRA LABS® offers Pre and Post Test Genetic Counseling

Personalized Genetic expertise for your patients

PRE-TEST CONSULTATION
Talk with a genetic expert by phone or video once your test is ordered. We review personal and family health history, answer any questions you may have, and determine if you meet eligibility for test authorization.

RESULTS CONSULTATION
Review test results with a genetic counselor or medical geneticist. During this phone or video session, you are provided an opportunity to discuss the results of the test and their implications.
With a focus on women’s health, ASPIRA LABS® offers both comprehensive and targeted genetic testing options.

### ASPIRA GenetiX Testing Options

**BRCA1 & BRCA2**
- **Genes Tested:** BRCA1, BRCA2

**Patient Considerations for Testing:**
- For the female relatives of women with breast or ovarian cancer.
- BRCA1/BRCA2 gene testing is indicated for women with a personal or family history of breast and/or ovarian cancer.
- BRCA1, BRCA2, and if negative for a Pathogenic or Likely Pathogenic variant, reflex to Targeted Breast & Ovarian 24.

**BRCA1 & BRCA2 Reflex to Targeted Breast and Ovarian 24**
- **Genes Tested:** BRCA1, BRCA2, and if negative for Pathogenic or Likely Pathogenic variants, reflex to Targeted Breast & Ovarian 24.

**BRCA1 & BRCA2 Reflex to Comprehensive Breast, Ovarian and Endometrial 34**
- **Genes Tested:** BRCA1, BRCA2, and if negative for Pathogenic or Likely Pathogenic variants, reflex to Comprehensive Breast, Ovarian and Endometrial 34.

**Targeted Breast & Ovarian 24 Gene Panel**
- **Genes Tested:** ATM, BRCA1, BRCA2, CHEK2, CDH1, CDKN2A/B, DCC, EPCAM, FGFR4, MSH2 (mono/biallelic), MLH1, MRE11A, NBN, NF1, PALB2, PTEN, RAD50, RAD51C, RAD51D, RAD54L, RAD55, RAD57, RAD60, SMARC1, SMARC2, STK11, TP53, WT1

**Patient Considerations for Testing:**
- BRCA1/BRCA2, the genes in the targeted panel include >25% of hereditary breast or ovarian cancer cases associated with these genes. This panel includes the genes in the Targeted Breast & Ovarian 24 plus Lynch Syndrome.

**Comprehensive Breast, Ovarian, and Endometrial 34 Gene Panel**
- **Genes Tested:** ATM, BRCA1, BRCA2, CHEK2, CDH1, CDKN2A/B, DCC, EPCAM, FGFR4, MSH2 (mono/biallelic), MLH1, MRE11A, NBN, NF1, PALB2, PTEN, RAD50, RAD51C, RAD51D, RAD54L, RAD55, RAD57, RAD60, SMARC1, SMARC2, STK11, TP53, WT1

**Patient Considerations for Testing:**
- BRCA1/BRCA2, the genes in the comprehensive panel include >50% of hereditary breast or ovarian cancer cases associated with these genes. This panel includes the genes in the Comprehensive Breast, Ovarian, and Endometrial 34 plus Lynch Syndrome.

**Lifeline risk of Ovarian cancer increases from 1.3% to 44% when a BRCA1 mutation is identified.**

*4 Reasons Why Your Patient Should Be Tested*
1. Identify risks for hereditary reproductive cancer health conditions
2. Help inform the management plan for patients with a positive test result
3. Determine if close family members (children, siblings, and/or parents) may also be at risk

**Gynecologic Cancer Predisposition Syndromes Covered**

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<th>BRCA1/BRCA2</th>
<th>Lynch Syndrome</th>
<th>Ovarian</th>
<th>Breast</th>
<th>Endometrial</th>
<th>Colorectal</th>
<th>New Evidence</th>
<th>Gene Panel</th>
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In addition to 19-28% endometrial cancer risk, women with germline mutations in the PTEN gene have up to a 50% risk of breast cancer and a 3-10% risk of thyroid cancer.