Uncovering Risk to Optimize Treatment, with a **Gynecologic Focus**

ASPIRA GENETIX
BY ASPIRA LABS

ASPIRA
WOMEN'S HEALTH™

Genetic Testing Options to Provide Personalized Care

With a focus on womens health, ASPIRA LABS® offers **both comprehensive and targeted genetic testing options** for Hereditary Breast and Ovarian cancer to help guide medical management for your patient.

Red flags for hereditary cancers can include early onset of cancer before age 50, more than one primary cancer in a single person, or cancers within a family.

Transforming Women's Health with an Innovative Solution

New Approach in the Genetic Testing Market

- Over 10 years of expertise in Ovarian Cancer Risk assessment and Pelvic mass management
- Cutting edge bioinformatics for analysis and data management
- Experienced in genetic reporting and quality focused patient care
- Laboratory Research focused on innovation and discovery

Highlights of Coverage & Analysis

ASPIRA GENETIX leverages whole gene sequencing technology (coding regions and adjacent intronic/splice regions) with >99% bases covered by at least 20x.

High Quality Genetic Testing

Coverage*
99.9% at 50x

Turn around time
2 - 3 weeks

Sample Requirements
+ Blood (1 - 4mL EDTA, lavender top tube)
+ Saliva

Deletion & Duplication
≥ 1 exon resolution

Superior to technology to analyze complex regions

+ Typical coverage for coding sequences: 99% at > 50x
+ Deletion and duplication analysis with single exon resolution for all genes
+ *BRCA2*: Portuguese Founder Mutation
+ *MSH2*: Boland Inversion (exon 1-7 inversion)
+ *PTEN*: Promoter region
+ *TP53*: Promoter region
+ *EPCAM* promoter del/dup detection
+ *PMS2*: Analysis includes exons 1-5 and 12-15

Lifetime risk of Ovarian Cancer increases from 1.3% to 44% when a BRCA1 mutation is identified.[1]

* Full gene sequencing and deletion/duplication, including detection of MSH2 inversion, BRCA2 Alu variant, and PMS2 (including exons 1-5 and 12.15), as well as sequencing of noncoding regions for selected genes.

1. https://www.cancer.gov/about-cancer/causes-prevention/genetics/brca-fact-sheet#q2

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ASPIRA GENETIX Testing Options

BRCA1 & BRCA2

Genes Tested:
BRCA1, BRCA2

Patient Considerations for Testing:
BRCA1 & *BRCA2* are well studied genes that account for 5-15% of hereditary breast or ovarian cancer cases (includes *BRCA2* Portuguese Mutation).

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 and Ovarian 24

Patient Considerations for Testing:
In patients without variants identified in *BRCA1* & *BRCA2*, the genes included in the 24 gene targeted panel will be analyzed to uncover potential genetic risk.

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

Patient Considerations for Testing:
In patients without variants identified in *BRCA1* & *BRCA2*, the genes included in the comprehensive 34 gene panel will be analyzed to uncover potential genetic risk with additional genes associated with female reproductive cancers.

Targeted Breast & Ovarian 24 Gene Panel

Genes Tested:
ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1,CHEK2, DICER, EPCAM, MLH1, MSH2, (mono/biallelic), MSH6, MRE11A, NBN, NFI, PALB2,PMS2, PTEN, RAD50, RAD51C, RAD51D, SMARCA4, STK11, TP53

Patient Considerations for Testing:
Including *BRCA1* & *BRCA2*, the genes in the targeted panel include elevated, moderate to high risk genes with management treatment plans as recommended by NCCN*, including syndromic associated genes with elevated risk of HBOC (such as Lynch Syndrome). Patients with a family history of breast and/or ovarian cancer without a known family history of *BRCA1* or *BRCA2* mutations may benefit from this option.

**Jan. 2019 NCCN ovarian cancer guidelines*

Comprehensive Breast, Ovarian and Endometrial 34 Gene Panel

Genes Tested:
ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1,CHEK2, DICER1, EPCAM, FANCC, FANCM, MLH1, MRE11(A), MSH2, (mono/biallelic), MSH6, MUTYH, NBN, NF1, NTHL1 (mono/biallelic),PALB2, POLE, POLD1,PMS2, PTEN, RAD50,RAD51C, RAD51D, SDHB, SDHD, SMARCA4, STK11,TP53, WRN, XRCC2

Patient Considerations for Testing:
Including *BRCA1* & *BRCA2*, the genes in the comprehensive panel includes genes with strong associations with hereditary breast and ovarian cancer as well as newer evidence genes and some genes associated with uterine & endometrial cancer risk. Patients with a family history of breast, ovarian or endometrial cancer without a known family history of and/or ovarian cancer without a known family history of *BRCA1* or *BRCA2* mutations may benefit from this option.

4 Reasons Why Your Patient Should be Tested

1. Establish or confirm a diagnosis
2. Identify risks for women's reproductive cancer health conditions
3. Help inform the management plan for patients with a positive test result
4. Determine if close family members (children, siblings, and/or parents) may also be at risk

Gynecologic Cancer Predisposition Syndromes Covered

	BRCA1/ BRCA2	**Lynch Syndrome** *(MLH1, MSH2, MSH6, PMS2, EPCAM)*	**Peutz-Jeghers Syndrome** *(STK11)*	**Li-Fraumeni Syndrome** *(TP53)*	**Cowden Syndrome** *(PTEN)*	**New Evidence Genes** *(FANCC, MRE11, NFI, NTHL1)*
BRCA1/BRCA2	✓					
Targeted Breast and Ovarian 24	✓	✓	✓	✓	✓	
Comprehensive Breast, Ovarian and Endometrial 34	✓	✓	✓	✓	✓	✓

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 3–10% risk of thyroid cancer.

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.[1]

Evaluation of the likelihood of a patient having a gynecologic cancer predisposition syndromes enables physicians to **provide individualized assessments of cancer risk.**[2]

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.

1. ACOG Practice Bulletin Number 182, September 2017

2. Lancaster, Johnathan M. et al. Society of Gynecologic Oncology statement on risk assessment for inherited gynecologic cancer predispositions. *Gynecologic Oncology*, Volume 136, Issue 1, 3 - 7

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