HEREDITARY CANCER TESTING
WHAT YOU AND YOUR PATIENTS WANT AND DESERVE

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DISCLOSURES

THIS SPEAKER HAS RELEVANT FINANCIAL RELATIONSHIPS WITH THE FOLLOWING COMMERCIAL INTERESTS:

• NATIONAL PROCTOR AND SPEAKER FOR INTUITIVE SURGICAL
• NATIONAL SPEAKER FOR MYRIAD GENETICS
• HUSBAND, FATHER OF 4 CHILDREN ALL UNDER AGE 11
• MINIMAL ASSETS, ALWAYS LOOKING FOR MORE
AT THE CONCLUSION OF THIS PRESENTATION, PARTICIPANTS SHOULD BE ABLE TO:

- Utilize Cancer Family History (CFHx) to optimally manage all patients
- Stratify patients by risk categories to help determine appropriate management and screening recommendations
- Integrate a Hereditary Cancer Risk Assessment (HCRA) protocol into your practice
- Discuss how technology impacts patient care
COMMON GAPS IN PATIENT CARE

• Not testing the same day
• I’m already doing a great job
• Time consuming
• Patient compliance - fear
• Inadequate documentation
• Miscommunication, refer out to GC
• Lack of coordinated follow-up
• Inadequate informed consent
• Not my responsibility
• Testing the index patient
• Cost of testing and the cost of medicine
Risk Stratification

• Cancer family history alone can help you optimize management.

• If your patient is positive for a syndrome, management will be different. Even a negative result will impact medical management.

Genetic testing is the only way to stratify risk between these two groups and find those at highest risk for cancer.
Hereditary Cancer Syndromes

**Hereditary Breast & Ovarian Cancer (HBOC)**

- **General Population**
  - Breast Cancer by Age 70: 7.3% (7.3%)<br>  - Colon Cancer by Age 70: 15-40% (15-40%)<br>  - Endometrial Cancer by Age 70: 4-11% (4-11%)

- **Familial Risk**
  - Breast Cancer by Age 70: 15-40% (15-40%)<br>  - Colon Cancer by Age 70: 4-11% (4-11%)<br>  - Endometrial Cancer by Age 70: 4-11% (4-11%)

- **Hereditary Risk**
  - Breast Cancer by Age 70: up to 87% (up to 87%)<br>  - Colon Cancer by Age 70: up to 44% (up to 44%)

**Lynch Syndrome**

- **General Population**
  - Breast Cancer by Age 70: 2% (2%)<br>  - Colon Cancer by Age 70: 4-20% (4-20%)

- **Familial Risk**
  - Breast Cancer by Age 70: 2% (2%)<br>  - Colon Cancer by Age 70: 4-11% (4-11%)

- **Hereditary Risk**
  - Breast Cancer by Age 70: up to 82% (up to 82%)<br>  - Colon Cancer by Age 70: up to 71% (up to 71%)
The Clinical Dilemma

**Multiple genes** can be associated with increased risk of a single cancer

**Multiple cancer risks** can be associated with a single gene

Assessment that is too narrow can lead to a false sense of security and patient mismanagement.
CLINICAL CASE #1

• A 39-year-old patient has AUB and had a failed ablation. Pt would like definitive care.

• Patients family history
  – Mom breast cancer age 52
  – Sister ovarian cancer age 29
  – Dad colon cancer age 49
  – Uncle “belly” cancer unknown age

• What test would you order, if any, prior to boarding her for surgery?

• When boarding this patient for surgery would you take her tubes and ovaries along with the uterus?

• Is there any testing that would change your decision making process?

• Would this information impact your medical/surgical decision making?
• Patient came in for a Well Woman Exam
• Patient family history:
  – Ashkenazi Jewish
  – Mom breast cancer, 55 yrs, BRCA negative
  – Aunt breast cancer, 65 yrs
  – Grandma breast cancer, 50s
  – Dad colon cancer, early 50s
• Patient receiving high risk breast protocol already
• Panel test finds patient is positive for Chek2
  • Increased risk of colon cancer and children now need testing at appropriate age
• ACA covers 94% of testing for patients who qualify for BRCA testing
“Evaluating a patient’s risk for hereditary breast and ovarian cancer syndrome (HBOC) should be a ROUTINE part of obstetric and gynecological practice”
Clinical Guidelines and Standardization of Practice to Improve Outcomes

“Protocols and checklists have been shown to reduce patient harm through improved standardization and communication...the use of checklists and protocols clearly has been demonstrated to improve outcomes and their use is strongly encouraged. Checklists and protocols should be incorporated into systems as a way to help practitioners provide the best evidence-based care to their patients.”
ACOG COMMITTEE OPINION 478 REAFFIRMED 2015

• “family history plays a very important role in assessing the risk of inherited medical conditions and single gene disorders...”

• “It is recommended that ALL women receive a family history evaluation as a screening tool for inherited risk. Family history information should be reviewed and updated regularly, especially when there are significant changes to family history.”
“A hereditary cancer risk assessment is the key to identifying patients and families who may be at increased risk of developing certain types of cancer. This assessment should be performed by obstetrician-gynecologists or other obstetric-gynecologic providers and should be updated regularly.”
ACOG'S POSITION ON REFERRALS TO GENETIC COUNSELORS

Committee Opinion 634, June 2015

• States OBGYNs can refer to a genetic counselor but does not state that an OBGYN should not order a genetic test.

July 23, 2015 ACOG Newsletter, A Message from President Mark DeFrancesco, MD, MBA

• “ACOG pushed back on a CMS proposal that would prohibit ob-gyns from conducting genetic counseling and would have required a go-ahead from an outside genetic counselor before the physician could order genetic tests.”
• “ACOG firmly opposes any attempt to restrict an ob-gyn’s scope of practice or put barriers between our Fellows and the care you provide to your patients.”
HALF THE TIME THE CLINICAL IMPRESSION IS INCORRECT
Technological advancement in hereditary cancer testing allows greater assurance of optimal patient management.

The Society of Gynecologic Oncology (SGO) and the National Comprehensive Cancer Network (NCCN) recognize the benefits of hereditary cancer panels:\textsuperscript{1,2}

- Cost Effective Approach
- Improved Efficiency
- Greater Assurance in Test Results

2. NCCN Guidelines version 2.2015: Genetic/Familial High-Risk Assessment: Breast and Ovarian
Risk Assessment for Lynch Syndrome and Hereditary Breast and Ovarian Cancer Syndrome

Patient Name:       Physician:  ... hereditary cancer (BRCA/Colaris) in the past? YES NO

Have you or any of your relatives been tested for hereditary cancer (BRCA/Colaris) in the past?

This is a screening tool for cancers that run in families. Please consider these family members when completing the form:

Mother/Father/Sister/Brother/Children = 1st Degree Relatives
Aunt/Uncle/Grandparent/Niece/Nephew = 2nd Degree Relatives  Cousin/Great Grandparent = 3rd Degree Relatives

Have you or any of your relatives been tested for hereditary cancer (BRCA/Colaris) in the past? YES NO

Patient's signature:  Date:

FOR OFFICE USE ONLY

Patient is appropriate for further risk assessment and/or genetic testing  Information given to patient to review  Follow-up appointment scheduled on  Patient offered genetic testing:  Accepted  OR  Declined  HCP Signature:  

Colonic and Uterine Cancer (Lynch Syndrome/Colaris)

SELF

YOUR RELATIONSHIP TO FAMILY MEMBER w/ CANCER

AGE AT DIAGNOSIS

MOTHER'S SIDE  FATHER'S SIDE

EXAMPLE: Two or more relatives with a Lynch syndrome cancer; one under age 50

Y  N

Have YOU been diagnosed with uterine (endometrial) or colorectal cancer before age 50

Y  N

Two or more relatives on the same side of the family w/ any of the following, one diagnosed before 50 (please circle):

- Colon, uterine/endometrial, ovarian, stomach, small bowel, brain, kidney/urinary tract, ureter and renal pelvis

Y  N

Three or more relatives on the same side of the family w/ any of the following diagnosed at any age (please circle):

- Colon, uterine/endometrial, ovarian, stomach, small bowel, brain, kidney/urinary tract, ureter and renal pelvis

Y  N

Family member has a known Lynch syndrome mutation

Breast and Ovarian Cancer (HBOC/BRACAnalysis)

SELF

YOUR RELATIONSHIP TO FAMILY MEMBER w/ CANCER

AGE AT DIAGNOSIS

MOTHER'S SIDE  FATHER'S SIDE

Y  N

Breast cancer at age 45 or younger (in self, first or second degree family members)

Y  N

Ovarian cancer at any age (in self, first or second degree family members)

Y  N

Two relatives on the same side of the family with breast cancer— with one under the age of 50

Y  N

Three relatives on the same side of the family with breast cancer at any age

Y  N

Multiple breast cancers in the same person (in the same breast or in both breasts)

Y  N

Male breast cancer at any age

Y  N

Ashkenazi Jewish ancestry with breast, ovarian or pancreatic cancer in the same person or on the same side of the family

Y  N

Pancreatic cancer with breast or ovarian cancer in the same person or on the same side of the family

Y  N

Triple Negative breast cancer under age 60 (ER, PR and Her2 negative receptor status)

Y  N

A family member with a known BRCA mutation

Is there any other cancer in you or any family members not listed above (provide site, relationship and age):

Patient's signature:  Date:  

This is a screening tool for cancers that run in families. Please consider these family members when completing the form:
Informed Consent or Refusal

If patient meets testing criteria:

- Discuss testing with patients just as you do with other common diagnostic tests such as a colposcopy
- Emphasize the need for a diagnostic test result in order to manage the patient optimally
- Obtain and document patient’s consent or refusal
WHY I FEEL CONFIDENT WITH MY PATIENTS’ RESULTS

• I test with a lab that:
  • Has experience finding mutations in DNA
  • Can accurately classify mutations
  • Reports updated classification data
  • Publishes data to support its claims
Family history may be the only symptom.
MY FOUR CHILDREN