Who should consider genetic counseling?

Personal and/or family history of cancer suggestive of a hereditary syndrome include:
- Breast cancer 45 or younger
- Premenopausal breast cancer
- Male breast cancer
- Ovarian cancer
- Multiple relatives on the same side of the family with breast, ovarian, prostate, colon or pancreatic cancers
- Ashkenazi Jewish heritage
- Rare cancers
- Young age of diagnosis
- Relative with positive results for any hereditary cancer gene

DEAR MEDICAL PROFESSIONAL

In January of 2017, EWBC began wide-scale risk assessment for our patients. Determining an individual’s lifetime risk of breast cancer is imperative in implementing an appropriate management plan. We chose CRA (formerly Hughes Risk App) as it is an up-to-date compilation of current risk assessment models.

Our patients complete a health history questionnaire which allows the software to calculate both the patient’s lifetime risk of breast cancer as well as their individual likelihood of a genetic mutation. Any patient with a personal or family history of cancer meeting NCCN guidelines or a 5% risk or greater of a mutation is eligible for genetic counseling and possible testing. Referring physicians receive a report which includes all appropriate risk scores.

Each of the three most highly recommended risk models (BRCAPRO, Claus, Tyrer-Cuzick) take into account different risk factors and therefore, can calculate slightly different scores. We find Tyrer-Cuzick to be the most comprehensive for the majority of our patients as it most accurately includes family history of breast and ovarian cancers. Additionally, if the patient’s personal or family history changes between appointments, the risk calculation score will be recalculated to include these updates. The complex and highly variable nature of the risk calculations is why we elected to adopt the most current software. Overall, the results are accurate and highly reproducible if utilizing the most up to date risk models.

We hope this newsletter will give you some insight into our cancer risk assessment and genetic counseling program and answers some questions you may have.

EWBC Cancer Risk Assessment & Genetic Counseling Program (585) 758-7041
### Risk Assessment Models

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<tr>
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<tbody>
<tr>
<td>Body mass index</td>
<td>No</td>
<td>Yes</td>
<td>No</td>
<td>Yes</td>
</tr>
<tr>
<td>Age at menarche</td>
<td>Yes</td>
<td>No</td>
<td>No</td>
<td>Yes</td>
</tr>
<tr>
<td>Age at first live birth</td>
<td>Yes</td>
<td>No</td>
<td>No</td>
<td>Yes</td>
</tr>
<tr>
<td>Age at menopause</td>
<td>No</td>
<td>No</td>
<td>No</td>
<td>Yes</td>
</tr>
<tr>
<td>HRT use</td>
<td>No</td>
<td>No</td>
<td>No</td>
<td>Yes</td>
</tr>
<tr>
<td>Breast biopsies</td>
<td>Yes</td>
<td>No</td>
<td>No</td>
<td>Yes</td>
</tr>
<tr>
<td>ADH</td>
<td>Yes</td>
<td>No</td>
<td>No</td>
<td>Yes</td>
</tr>
<tr>
<td>LCIS</td>
<td>No</td>
<td>No</td>
<td>No</td>
<td>Yes</td>
</tr>
<tr>
<td>Breast density</td>
<td>No</td>
<td>No</td>
<td>No</td>
<td>No</td>
</tr>
<tr>
<td>First-degree relative</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>Second-degree relative</td>
<td>No</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>Age of onset</td>
<td>No</td>
<td>Yes</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>Bilateral cancer</td>
<td>No</td>
<td>No</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>Ovarian cancer</td>
<td>No</td>
<td>No</td>
<td>Yes</td>
<td>Yes</td>
</tr>
<tr>
<td>Male breast cancer</td>
<td>No</td>
<td>No</td>
<td>Yes</td>
<td>Yes</td>
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</table>

### How we use the Tyrer-Cuzick Model

**Factors Considered**
- Age, height, weight
- Jewish ethnicity
- Age at menarche, menopause, and age at first pregnancy or nulliparity
- HRT use
- History of hyperplasia (+/- atypia) and LCIS
- Extended paternal and maternal family history of both breast and ovarian cancer, including ages of onset (male breast cancer included in v7 and v8)
- Genetic test results
- Density (v8 only)

**Output**
- 10 year breast cancer risk
- Lifetime breast cancer risk
- Probability of BRCA1/2 gene mutation

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### Additional Tools

**BCSC Risk Calculator:** [http://tools.bcs-scc.org](http://tools.bcs-scc.org)

**BOADICEA***: [http://ccge.medschl.cam.ac.uk/boadicea](http://ccge.medschl.cam.ac.uk/boadicea)

* Endorsed by ACS for identifying breast MRI screening candidates

### References

**GAIL:** *J Natl Cancer Inst* 1995;87(22):1681-1685.

**Claus***: *Cancer* 1994;73:643-51.

**IBIS (Tyrer-Cuzick)***: *Stat Med* 2004;23(7):1111-1130.


**BRCAPRO***: *Cancer* 2002;94(2):305-313.


**All SCREENING GUIDELINES are individualized to a younger age if a relative has been diagnosed before age 30. Please see gene specific guidelines included with patient results, as the age for starting MRI and/or Mammography differs by gene.**  

*e.g.: BRCA1 and BRCA2 – age 25 | PALB2 – age 30 | ATM and CHEK2 – age 40*

<table>
<thead>
<tr>
<th>NCCN</th>
<th>INTERVENTION</th>
<th>GENES</th>
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<tbody>
<tr>
<td>Intervention Warranted Based on gene and/or Risk Level</td>
<td>Recommend Breast MRI (<em>20% lifetime risk</em>)</td>
<td>ATM, BRCA1, BRCA2, CDH1, CHEK2, PALB2, PTEN, STK11, TP53</td>
</tr>
<tr>
<td></td>
<td>Discuss Option of Risk-Reducing Mastectomy</td>
<td>BRCA1, BRCA2, CDH1, PTEN, TP53, PALB2</td>
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<tr>
<td></td>
<td>Recommend/Consider Risk-Reducing BSO</td>
<td>BRCA1, BRCA2, MLH1, MLH2, MSH6, PMS2, EPCAM, BRIP1, RAD51C, RAD51D</td>
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</table>

**FOR YOUR PATIENTS**

**GENETIC TESTING & BREAST CANCER COMMUNITY LECTURES**  
*presented by Jessica Salamone, ScM, CGC*

Main office in Brighton, 170 Sawgrass Drive, Rochester, NY 14620

**Mondays at 5:30pm:**
June 12, July 10, Aug 14, Oct 10 (Tuesday), Nov 13, Dec 11

Please sign up at (585) 442-2190, ext 6070

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**FOR YOU & YOUR STAFF**

**GENETICS LUNCH & LEARN FOR MEDICAL PROFESSIONALS**  
*presented by Jessica Salamone, ScM, CGC*

Please call EWBC (585) 442-2190, ext 6070
For brochures and scripts Please call (585) 758-7041

**REFERRALS or QUESTIONS**
Jessica Salamone, ScM, CGC
jsalamone@ewbc.com or (585) 758-7041
A PANEL APPROACH MAY CHANGE MEDICAL MANAGEMENT

*even if it includes genes not associated with breast cancer*

- **3.1%** Other high-risk genes associated with breast cancer (PTEN, TP53, CDH1 and STK11)
- **10.3%** Genes not associated with breast cancer (MLH1, MSH2, MSH6, PMS2, EPCAM, APC, MUTYH, CDKN2A, RAD51D, RAD51C and SMAD4)
- **48.7%** Hereditary breast and ovarian cancer syndrome-specific genes (BRCA1 and BRCA2)
- **37.9%** Moderate-risk genes associated with breast cancer (PALB2, CHEK2, ATM, BRIP1*, BARD1 and NBN)

*Evolving data suggests that BRIP may not be associated with significantly increased breast cancer risk.