

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.

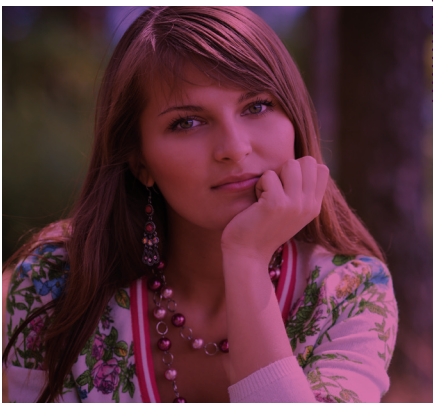
Determining an individual's lifetime risk of breast cancer is imperative in implementing an appropriate management plan. We chose CRA (formerly Hughes Risk App) as it is an up-to-date compilation of current risk assessment models.

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

	GAIL <i>www.cancer.gov/bcrisktool</i>	CLAUS* <i>iPhone/iPad app: BRisk</i>	BRCAPRO*	IBIS TYRER-CUZICK v.8* <i>http://www.ems-trials.org/riskevaluator/</i>
Body mass index	No	No	No	Yes
Age at menarche	Yes	No	No	Yes
Age at first live birth	Yes	No	No	Yes
Age at menopause	No	No	No	Yes
HRT use	No	No	No	Yes
Breast biopsies	Yes	No	No	Yes
ADH	Yes	No	No	Yes
LCIS	No	No	No	Yes
Breast density	No	No	No	Yes
First-degree relative	Yes	Yes	Yes	Yes
Second-degree relative	No	Yes	Yes	Yes
Age of onset	No	Yes	Yes	Yes
Bilateral cancer	No	No	Yes	Yes
Ovarian cancer	No	No	Yes	Yes
Male breast cancer	No	No	Yes	Yes

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



Additional Tools

BCSC Risk Calculator:

<http://tools.bscsc-scc.org>

BOADICEA*:

<http://ccge.medschl.cam.ac.uk/boadicea>

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.

BCSC Risk Calculator: *J Natl Cancer Inst* 2015;107(5):dju397.

BOADICEA*: *Br J Cancer* 2004;91(8):1580-1590.

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

J Clin Oncol 2002;20(11):2701-2712.

JNCI 2002;94(11):844-851.

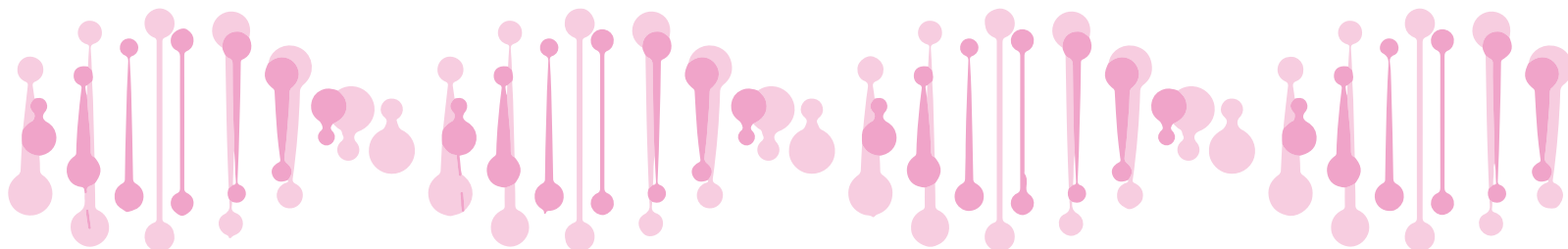
* Endorsed by ACS for identifying breast MRI screening candidates

MANAGEMENT OPTIONS

NCCN Guidelines (NATIONAL COMPREHENSIVE CANCER NETWORK)

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

*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*



Presentations

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

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

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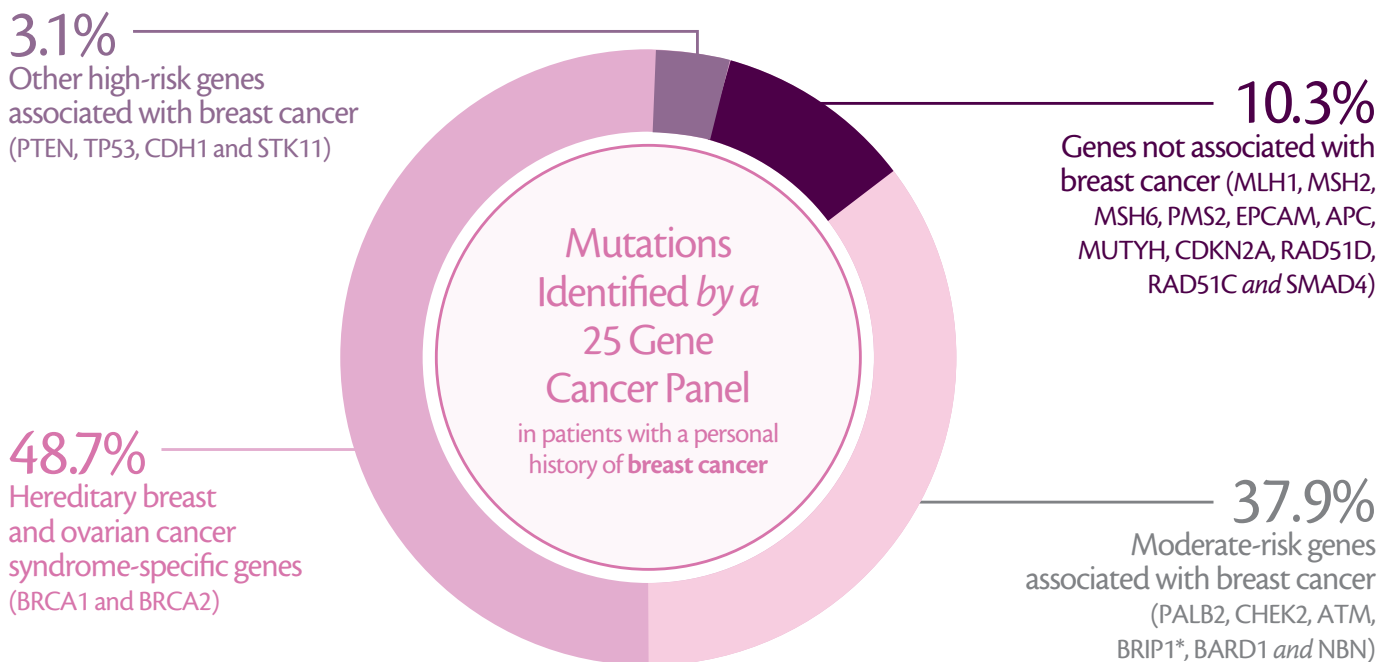


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A PANEL APPROACH MAY CHANGE MEDICAL MANAGEMENT *even if it includes genes not associated with breast cancer*



SOURCE: Sharma et al. Spectrum of Mutations Identified in a 25-gene Hereditary Cancer Panel for Patients with Breast Cancer. Presented at ESHG 2015

*Evolving data suggests that BRIP1 may not be associated with significantly increased breast cancer risk.