Female Breast Cancer — **BRCA1 and BRCA2 Genes**

Women with a mutation in the *BRCA1* gene have an increased lifetime risk of developing breast cancer of approximately 72 percent by age 80 (as compared to the general population lifetime risk of approximately 12 percent). Breast cancer tends to occur at a younger age in *BRCA1* carriers than the general population, with a peak incidence between 41 and 50 years.

Women with a mutation in the *BRCA2* gene have an increased lifetime risk of developing breast cancer of approximately 69 percent by age 80 (as compared to the general population lifetime risk of approximately 12 percent). Breast cancer tends to occur at a slightly younger age in *BRCA2* carriers than the general population, with a peak incidence between 51 and 60 years.

Management of breast cancer risk includes screening to maximize early detection of breast cancer, and may also include medications to reduce risk or risk-reducing surgery.

Screening for breast cancer may include a physical exam, mammogram, and MRI.

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A **mammogram** is an X-ray of the breast that can detect small abnormalities that cannot be felt on clinical exam. 3D mammograms are routinely used as they detect more breast cancers than traditional 2D mammograms. The radiation dose from mammograms is low, and the benefit of detecting breast cancer early outweighs the small risk of radiation.

A breast **MRI** uses a magnet to create images of the breasts and surrounding tissues. A breast MRI requires the use of contrast injected in the veins at the time of the study.

### Screening—Recommended Schedule

- Starting at age 25, clinical breast examination by a healthcare provider every 6-12 months.
- At ages 25-29, annual breast MRI.
- At ages 30 and older, annual mammogram in addition to annual breast MRI, staggered six months apart so there is not a full year between imaging studies.

**NOTE:** High-risk screening can be combined with risk-reducing medications.

### Risk-reducing Medications

Chemoprevention is an option for high-risk women. It involves taking medication(s) to try to lower the risk of developing breast cancer. Chemoprevention medications block the effects of estrogen and have been shown to decrease the risk of developing breast cancer in a high-risk population—some studies have shown a decrease in risk of 50 percent. However, the data on chemoprevention in women with **BRCA1** and **BRCA2** gene mutations is limited.

Examples of these medications include tamoxifen, raloxifene, and aromatase inhibitors. Our high-risk and breast cancer specialists will determine the most appropriate medication(s) and discuss specific side effects.
Risk-reducing Surgery

Bilateral prophylactic (preventive) mastectomy is the surgical removal of both breasts. It is the most effective way of reducing breast cancer risk in women with BRCA1 or BRCA2 gene mutations. This surgery reduces breast cancer risk by approximately 90 percent but does not eliminate it completely. You will be scheduled to see a breast surgeon who will discuss the details of this with you.

Prophylactic mastectomies can be performed in conjunction with reconstruction, done at the same time by a plastic surgeon. Reconstruction is an individual choice, and patients have the option to pursue if they would like. If considering a bilateral mastectomy, you may want to meet with a plastic surgeon to discuss options for reconstruction to help you make a decision that is right for you.

Male Breast Cancer — BRCA1 and BRCA2 Genes

Men with a mutation in the BRCA1 gene have an increased lifetime risk of developing breast cancer of approximately 1-2 percent by age 80 (as compared to 1 in 1,000, or 0.001 percent, for average-risk men). Men with a mutation in the BRCA2 gene have an increased lifetime risk of developing breast cancer of approximately 7 percent by age 80.

Men with a BRCA1 or BRCA2 gene mutation should begin breast self-exams and have an annual clinical breast exam with a specialist starting at age 35. Mammogram screening in men with BRCA1 or BRCA2 gene mutations should be done on an individualized basis.

Men with a BRCA1 or BRCA2 gene mutation have a significantly lower risk of developing breast cancer than women with a BRCA1 or BRCA2 gene mutation. Thus, prophylactic mastectomies are not typically pursued but can be considered on an individual basis.

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Breast Oncology Recommendations (cont.)

Our collaborative team of physicians and specialists works with you to develop a personalized breast cancer risk program that lets you choose the options that are best for you.

Mount Sinai’s Comprehensive BRCA Program for men and women who have mutations in the BRCA1 or BRCA2 gene provides expert guidance to ensure that you receive appropriate cancer screenings, monitoring, and treatment, as needed.

For information and appointments, call 877-309-BRCA (2722).

www.mountsinai.org/care/cancer/about/brca-program