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Hereditary Breast Cancer

WHAT YOU CAN – AND CAN'T – LEARN FROM GENETIC TESTING

Post-Mastectomy Breast Reconstruction Specialist Dr. Constance M Chen Answers Questions and Offers Tips

The most significant risk factors for breast cancer are being a woman and growing older. Among women in the general population, about one in eight (12%) will develop breast cancer over the course of her lifetime. However, among women who inherit a specific mutation in the BRCA1 or BRCA2 gene, the risk is significantly higher.

“Genetic testing can identify women who carry the harmful mutations,” says plastic surgeon and breast reconstruction specialist Dr. Constance M. Chen, “but these mutations are relatively rare in the general population and not every woman needs testing. It’s important to understand what we can and cannot determine from genetic testing, which women should have the test, and what can be done to reduce the risk for those who have inherited a mutated gene.”

What are BRCA genes? How do they affect breast cancer risk? The BRCA1 and BRCA2 genes produce proteins that help suppress tumors. These proteins play a role in repairing damaged DNA and maintaining the genetic stability of cells. When the genes mutate or are altered so they fail to produce the proteins or don't function properly, damaged DNA may not be repaired and cells are more likely to undergo further changes that can cause cancer. Specific inherited mutations in the BRCA1 and BRCA2 genes are associated with increased risk of breast and ovarian cancer and play a role in other cancers as well, such as pancreatic cancer, prostate cancer, and melanoma. The child of a parent whose mother or father carries the mutated gene has a one in two chance of inheriting it. Estimates are that 72% of women who inherit the BRCA1 mutation and 69% of those who inherit the BRCA2 mutation will develop breast cancer by the age of 80. In some cases, the risk for developing breast cancer can be over 86%.

Who should be tested for BRCA mutations?

“Compared to 12% of women overall who will have breast cancer, the increased risk associated with BRCA mutations is very substantial,” says Dr. Chen. “Given the risk, it’s natural to ask if all women should be tested. However, only about 1 in 400 people in the United States carry a BRCA1 or BRCA2 mutation. Thus, only people whose personal or family history is associated with greater likelihood of carrying the mutation are typically tested.” Indicators of a possible genetic mutation include a family history of breast or ovarian cancer, breast cancer diagnosed before age 50, male breast cancer, and cancer in both breasts. An additional risk factor is ethnicity; the BRCA mutation has a higher prevalence in women of Ashkenazi Jewish, Norwegian, Dutch, and Icelandic descent.

If I've already had breast cancer, is there any reason for me to be tested? Yes. If your personal or family history or ethnicity point to the likelihood of high risk of hereditary breast cancer, the results of genetic testing can help you be aware of your risk for other cancers, like ovarian cancer, and are important in helping other members of your family assess their risk.

If I test negative for harmful BRCA mutations, am I home free? Can I stop worrying about breast cancer?

“If only it were so,” says Dr. Chen. “About 90% of women who have breast cancer have no family history of the disease and no known genetic mutation to account for it. In the vast majority of cases, cancer is caused by spontaneous mutation, the aging process, or environmental factors, so a negative test for BRCA mutation is no guarantee that you will not develop breast cancer. All women should continue to have regular breast exams and mammograms.”

If I test positive for a harmful BRCA mutation, does that mean I will get cancer? What should I do?

A positive result significantly increases the risk of developing breast cancer. Many women in this position explore options for reducing their risk, the most effective of which is bilateral prophylactic mastectomy – the preventive removal of both breasts. According to the National Cancer Institute, women with harmful BRCA mutations who undergo prophylactic mastectomy can reduce their risk of developing breast cancer by 95%. “The risk reduction statistics are compelling,” says Dr. Chen, “but the decision to undergo prophylactic mastectomy is a difficult one. Those who are faced with this decision must weigh various medical and personal factors, including the breast reconstruction options that are available to them.”

Are there any other gene mutations that I should be aware of?

There are other genetic mutations like CHEK2, Lynch syndrome, PALB2, ATM, BRIP1, and others, which can also increase the risk of breast cancer. Talk with your doctor or genetic counselor about your individual circumstance. “Testing for BRCA and other genetic mutations has come a long way in the twenty or so years that it has been available,” Dr. Chen concludes. “In combination with genetic counseling, it provides valuable information that can guide key decisions for women and their families. However, the test can only help assess risk. Neither positive nor negative results provide definitive answers about who will or will not get cancer. Women and their doctors must consider test results along with other personal and medical factors to determine their best course of action.”

Constance M. Chen, MD, is a board-certified plastic surgeon with special expertise in the use of innovative natural techniques to optimize medical and cosmetic outcomes for women undergoing breast reconstruction. constancechenmd.com

Breast Implants Associated Anaplastic Large Cell Lymphoma

ALLERGAN SUSPENDS SALES OF ITS TEXTURED BREAST IMPLANTS IN EUROPE

Breast Reconstruction Specialist Dr. Constance M Chen with important information and tips for patients concerned about their breast implants

Allergan, one of the world's largest manufacturer of breast implants, suspended sales of its textured implants in Europe after the implants were denied renewed safety certificates in France because of concerns about BIA-ALCL. The French regulatory agency and a British association of plastic surgeons have recommended the use of smooth-surface implants in place of textured-surface products. Both European regulators and the U.S. FDA will hold meetings in 2019 to review further the safety of all breast implants but for now, textured implants remain approved for use in the U.S. "ALCL is a extremely rare cancer of the immune system, and questions have been raised about whether the incidence is higher in women undergoing breast reconstruction with breast implants," says Dr. Constance M Chen, board-certified plastic surgeon and breast reconstruction specialist. The incidence of ALCL in the breast is 3 cases of ALCL in 100 million women (0.000003%). Dr. Chen adds that "BIA-ALCL has been associated in women who have breast implants for both cosmetic and reconstructive reasons. Women who have breast implants for breast reconstruction due to mastectomy or for cosmetic breast augmentation are at equal risk for developing BIA-ALCL. The simple fact of having a breast implant - of any type, for any reason - in her body places a woman at higher risk of developing BIA-ALCL." Dr. Chen adds that the current literature estimates that BIA-ALCL may develop in 1 in 3,817 to 30,000 women with textured breast implants. "While attention has been focused on textured breast implants, BIA-ALCL has actually been found in all types of breast implants: textured, smooth, silicone, saline," says Dr. Chen. As of 30 September 2017, the FDA had received 414 medical device reports of BIA-ALCL, including the death of 9 patients. Only 272 of the 414 reports included information on the implant surface. Of these cases, BIA-ALCL was found associated with 242 implants with textured surfaces and 30 implants with smooth surfaces. With regard to implant filling, BIA-ALCL was found associated with 234 implants filled with silicone gel and 179 implants filled with saline. About half of the reported cases were diagnosed within 7-8 years of breast implantation.

The backstory: In 2011, the U.S. Food and Drug Administration (FDA) first noted that the incidence of ALCL seemed slightly higher in patients with breast implants, but it was still very rare even in this population with 60 cases of ALCL worldwide in 5-10 million women (0.0006-0.0012%). Dr. Chen adds, "that at the time, many healthcare professionals believed that the incidence of ALCL in women with breast implants might have even been overestimated because some of the data collected may have been duplicate cases." However, in 2016 data collection improved due to an increased attention to ALCL and breast implants, and the World Health Organization (WHO) designated breast implant-associated anaplastic large cell lymphoma (BIA-ALCL) as a T-cell lymphoma that can develop following breast implants. Several recent journal articles explore risk factors for developing BIA-ALCL, including the role of biofilm. Dr. Chen notes that "textured implants have significantly more surface area than smooth implants, which may explain why there is a higher incidence of BIA-ALCL in textured implants than smooth implants." However, BIA-ALCL has clearly been found associated with every type of breast implant.

Textured implants may be at slightly greater risk for ALCL than smooth implants, but findings are not conclusive. Dr. Chen points out that "this is important to note because many plastic surgeons have stopped using textured breast implants, but they continue using smooth breast implants. It is clearly not true that BIA-ALCL is limited to textured breast implants only. Since every type of breast implant has been associated with BIA-ALCL, it is a false sense of security to limit breast implant use to smooth breast implants. In reality, all types of breast implants can lead to BIA-ALCL."

Tips for Patients:

When ALCL occurs with breast implants, it is identified most frequently in patients undergoing implant revisions for late onset, persistent seroma (a fluid collection that develops around the implant), or other symptoms such as pain, lumps, swelling, or breast asymmetry. Dr. Chen advises that "any women experiencing these symptoms should see a plastic surgeon to be evaluated for BIA-ALCL." The FDA already recommends that women with silicone breast implants undergo breast MRI every 2-3 years to evaluate for silent rupture of her silicone breast implants. Dr. Chen advises that if a patient with breast implants feels a change in her breasts, breast imaging can be obtained to evaluate for seroma. If a seroma is found, an interventional radiologist or a skilled breast radiologist can aspirate the seroma fluid to test for CD30 markers and lymphoma. If the cytology is positive, the patient will need to be treated. Treatment of BIA-ALCL includes removal of the implant and capsule surrounding the implant and sometimes chemotherapy and radiation. If ALCL is diagnosed through imaging and testing of the fluid around the breast, treatment is generally the surgical removal of the implant and the entire capsule that surrounds it. In its early stages, all traces of cancer are generally removed by surgery. Further treatment is necessary only if all the cancer could not be removed or if the disease has spread to the lymph nodes or other parts of the body. Dr. Chen clarifies that the FDA does not currently recommend prophylactic breast implant removal in patients without symptoms or other abnormalities. At present, breast implants are still approved in the USA when used as labeled for breast augmentation or breast reconstruction, as the risk of ALCL is still considered to be small and inconclusive. All of this may soon change, however, as new and better data is collected on BIA-ALCL. "Women with breast implants who want to have their breast implants removed have options," says Dr. Chen. A woman facing breast reconstruction after mastectomy can consider autologous reconstruction, in which a new breast is created from her own tissue. If she is not a candidate for autologous reconstruction or if she prefers implants, she must be aware of the risk of BIA-ALCL. A woman who had breast implants for cosmetic breast augmentation can have her breast implants removed and undergo natural breast augmentation or accept a smaller size breast. Dr. Chen concludes, "even though breast implants of all types are still approved for use in the United States, any woman who has breast implants or is considering breast implants should be aware of BIA-ALCL so that she can be fully educated and consider the potential repercussions of breast implants on her health and well-being."

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