

Avoid Missed or Delayed Diagnosis by Being Aware of Risks for BRCA-Based Breast Cancer

Recent news coverage has brought BRCA gene-based breast cancer into the spotlight. Actress Angelina Jolie's decision to get a preventive double mastectomy after testing positive for the BRCA gene may cause patients to ask physicians if they are at risk. Physicians should be aware of the risk factors for BRCA gene-based cancer in order to identify those who need testing and to avoid delayed or missed diagnosis.

A recent malpractice case highlights the failure of missing an early diagnosis. A 33-year-old woman had two female relatives, including her mother, who had breast cancer in their forties. At 31, she began getting annual screening mammograms, which showed dense breasts. She complained of a small palpable mass. However, no mass was seen on a mammogram, and the diagnosis was fibrocystic changes. No additional tests were ordered. Within six months, the mass was enlarging, and she was diagnosed with infiltrating ductal cancer that had advanced from a Stage I to a Stage III. Based on her history, she should have been tested for the BRCA mutation and given various treatment options. Additionally, no ultrasounds or MRIs were done, which possibly could have detected the cancer at an earlier treatable stage.

A woman's risk of developing breast and/or ovarian cancer greatly increases if she inherits a BRCA1 or BRCA2 gene mutation. Widespread screening is not required because together these mutations account for only 5–10 percent of breast cancers. Those with the BRCA1 mutation have a 55–65 percent chance of developing breast cancer by age 70, and those with the BRCA2 mutation have a 45 percent chance. Women have about a 2 percent chance of getting ovarian cancer, but if they have a BRCA2 mutation, that risk increases to 40–60 percent.

Physicians should watch for the following BRCA mutation risk factors and discuss genetic testing with patients at risk:

- Maternal or paternal blood relatives with breast cancer diagnosed before the age of 50.
- Certain cancers in a patient's family, such as pancreatic, colon, or thyroid.
- Both breast and ovarian cancer in a patient's family, especially in one individual.
- Women in a patient's family with cancer in both breasts.
- Patient with Ashkenazi Jewish heritage.
- A male in the patient's family with breast cancer.
- Relative with BRCA1 or BRCA2 mutation.

If the patient does test positive for the BRCA mutation, it is essential to remind her that this does not indicate she will get cancer. Patients can reduce risks of cancer with prophylactic surgery, hormonal treatment, and lifestyle changes.

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